

Institute of Neuropathology, RWTH Aachen University Hospital

Publications 2026

Brenner D, Ponomarenko A, Petrut I, Beyrle S, Contardo M, Loss I, **Radke C**, Frank J, Zimmer E, Schlesner M, **Achenbach P**, Scheveneels W, Aly A, Nazlican H, Hesebeck-Brinkmann J, Oeckl P, Müller K, Siebert R, Böckers T, van Eijk K, Veldink J, Kleger A, Mulaw M, Andersen PM, Forsberg K, Weishaupt JH, Loghmani SB, Grehl T, van Damme P, **Weis J**, Catanese A. A rare missense variant impacting NEK1 kinase function is associated with ALS. *Acta Neuropathol Commun*. 2026 Jun 25;14(1):135. doi: 10.1186/s40478-026-02351-6. PMID: 42351313

Tripathi P, **Guo H**, **Yamoah A**, Mathur R, Doukas P, **Dresler A**, **Jesse CM**, Aronica E, Hermann A, Steinbusch H, **Brook GA**, **Weis J**, **Goswami A**. VAPB confers selective neuroprotection by driving autophagic degradation of pathogenic aggregates in ALS. *Acta Neuropathol Commun*. 2026 May 29;14(1):127. doi: 10.1186/s40478-026-02298-8. PMID: 42210413

Bock A, Schurig M, Willoughby M, Mirecki A, Seemann E, Lohachova K, **Katona I**, Mittag S, Liebmann L, Franzka P, Heidari Horestani M, Khundadze M, Mosler T, Louie T, de Visser M, Weterman MAJ, Kiehnopf M, Beetz C, Nietzsche S, Huber O, **Weis J**, Kessels MM, Bhaskara RM, Qualmann B, Đikić I, Hübner CA. REEP1 accumulation disrupts ER integrity and drives spinal motoneuron degeneration in distal hereditary motor neuropathy. *Adv Sci (Weinh)*. 13(2): e11483. 2026 doi: 10.1002/adv.202511483.

Zhang X, **Zajt KK**, **Palaz T**, **Wang L**, **Groß M**, Kraft F, **Weis J**, **Bremer J**. The chaperonin TRiC component Cct3 is required for axonal transport, myelination, and neuromuscular junction refinement. *Cell Death Dis*. 2026 Feb 12;17(1):221. doi: 10.1038/s41419-026-08465-y. PMID: 41680121

Dohrn MF, Pareyson D, Pisciotta C, Kölbel H, Roos A, Fabrizi GM, Cakar A, Parman Y, Züchner S, **Weis J**. Charcot-Marie-Tooth-SORD: insights into pathology and pathophysiology from a human nerve biopsy series. *Brain*. 2026 Mar 5; 149(3): e23-e26.1093/brain/awaf420. PMID: 41206017.

Jacob M, Kölbel H, Harrer P, Kopajtich R, Munot P, Achleitner MT, Badmann S, Brugger M, Brunet T, Bonne G, Codina M, Ebner L, Eshraghi P, Eyring K, Farhat AS, Feichtinger RG, Graf E, Marcé-Grau A, Hahn A, Houlden H, Karimiani EG, Manel V, Mayerhanser K, Nectoux J, Nelson I, Phadke R, Prokisch H, Sadeghian S, Saparov A, Schänzer A, Schara-Schmidt U, Schmidt J, Schuler R, Sewry C, Shariati G, Slanz S, Smirnov D, Sukenik-Halevy R, Tajsharghi H, Toosi MB, Trujillano L, **Weis J**, Wilson LC, Ben Yaou R, Zamani M, Zech M, Zschüntzsch J, Kornak U, Gómez- Andrés D, Maroofian R, Winkelmann J, Roos A, Distelmaier F, Mayr JA, Wagner M. Deciphering DST-associated disorders: biallelic variants affecting DST-b cause a congenital myopathy. *Brain*. 2026 Feb 7; 149(2): 653-667. doi: 10.1093/brain/awaf227. PMID: 40497796.

Publications 2025

van den Braak NWM, Kuehs S, Peschke GZ, Namer B, Lischka A, Eggermann K, Dumke C, Sudha Bhagavath Eswaran V, **Nikolin S**, **Weis J**, Schulz JB, Lampert A, Kurth I, Rolke R, Leipold E, Dohrn MF. Altered NaV1.9 channel activity in two Tyr66Ser variant carriers with small fiber dysfunction. *J Gen Physiol*. 2025 157(6): e202413691 doi: 10.1085/jgp.202413691

Bremer J, Nagel J, Zschüntzsch J, Zajt KK, **Palaz T**, Blank T, **Ikis A**, Fischer LA, Sensmeyer ASM, Wiechers L, Reichelt JJ, Hofmann KP, Wolf MJ, Leuchtenberger C, Tripathi P, Einer C, Zischka H, Rothermel U, Eck AL, Reimann R, Kana V, Rushing E, Aguzzi A, Prinz M, Liebetanz D, Odoardi F, Kuo

CC, **Weis J**, Kraft F, Schmidt J, Heikenwälder M. Mutual reinforcement of lymphotoxin-driven myositis and impaired autophagy in murine muscle. *Brain* 148(12): 4461-4481, 2025. PMID: 40853947.

Umathum V, König C, Bandorski D, Scheffer L, **Weis J**, Krämer HH, Allendörfer J, Schänzer A. Post-infectious autoimmune disorder involving the CNS and PNS following SARS-CoV-2 infection - a clinical-morphological case report. *Neurol Res Pract.* 2025 Oct 15;7(1): 76. doi: 10.1186/s42466-025-00436-6. PMID: 41094569; PMCID: PMC12529787.

Sofia Costa A, Pinho J, Reich A, Nikoubashman O, **Nolte K**, **Weis J**, Boy C, M Mottaghy F, B Schulz J, Reetz K. Iatrogenic cerebral amyloid angiopathy: two new cases and systematic review of case reports with neuropathological data. *Neurol Res Pract.* 2025 Sep 3;7(1): 63. doi: 10.1186/s42466-025-00423-x. PMID: 40903767; PMCID: PMC12409930.

Bahm J, Schäfer B, **Nolte K**, **Weis J**, Beier JP. Intraoperative assessment of nerve traction injury in obstetric brachial plexus palsy. *J Brachial Plex Peripher Nerve Inj.* 2025 May 2;20(1):e27-e30. doi: 10.1055/a-2572-2601. PMID: 40321364

Quade A, Lischka A, Albani S, Rossetti G, Hageb ZN, Rolke R, Kurth I, Katona IK, Eggermann K, Namer B, Lampert A, Dohrn MF, Schacht GM, **Weis J**, Häusler M. Genetic variants and clinical phenotyping in 39 pediatric patients with neuropathic pain. *Neuropediatrics.* 56(4): 249-258, 2025. PMID: 40294637.

Jütten K, Ort J, Kernbach JM, Meyer-Baese A, Meyer-Baese U, Hamou HA, Clusmann H, Wiesmann M, **Bremer J**, Koch H, Bak A, Ricklefs F, Drexler R, Heiland DH, Delev D. High peritumoral network connectedness in glioblastoma reveals a distinct epigenetic signature and is associated with decreased overall survival. *Neuro Oncol.* 2025 Oct 1; 27(10): 2564-2573. doi: 10.1093/neuonc/noaf101. PMID: 40230019

Achenbach P, **Altinova H**, **Brook GA**. Substrate topography as a powerful tool to modify glial cell biology and interactions. *Neural Regen Res.* 20(5): 1390-1391, 2025. PMID: 39075898

Vendredy L, De Winter V, Van Lent J, Orije J, Authier TDS, **Katona I**, Asselbergh B, Adriaenssens E, **Weis J**, Verhoye M, Timmerman V. RNA interference targeting small heat shock protein B8 failed to improve distal hereditary motor neuropathy in the mouse model. *J Gene Med.* 2025 Feb;27(2): e70013. doi: 10.1002/jgm.70013. PMID: 39972648

Schwarz D, Marois ML, Sturm V, Peters AS, Longuespée R, Helm D, Schneider M, Eichmüller B, Hidmark AS, Fischer M, Kender Z, Schwab C, Hausser I, **Weis J**, Dihlmann S, Böckler D, Bendszus M, Heiland S, Herzig S, Nawroth PP, Szendroedi J, Fleming T. Exploring structural and molecular features of sciatic nerve lesions in diabetic neuropathy: Unveiling pathogenic pathways and targets. *Diabetes.* 74(1): 65-74, 2025. PMID: 39418320.

Bremer J, Franco P, Menstell JA, **Tey S**, **Zajt KK**, **Popzhelyazkova K**, **Nolte K**, Schlegel J, Pedro M T, Osterloh A, Delev D, Hohenhaus M, Scholz C, Schnell O, Beck J, **Weis J**, Heiland DH. Spatially resolved transcriptomics of benign and malignant peripheral nerve sheath tumors. *Neuro Oncol.* 27(8):2073-2087, 2025. PMID: 39847441

Alves-Simões M, Teege L, Tomni C, Lürkens M, Schmidt A, Iseppon F, Millet Q, Kühs S, **Katona I**, **Weis J**, Heinemann SH, Hübner CA, Wood J, Leipold E, Kurth I, Haag N. NaV1.8/NaV1.9 double deletion mildly affects acute pain responses in mice. *Pain* 2025 Apr 1; 166(4):773-792. PMID: 39382328

Na CH, Ridwan H, Neuloh G, Schubert GA, **Nolte K**, Prescher A, Clusmann H, Blume C. Arachnoid web-a rare but surgically effectively treatable cause of spinal cord compression and syringomyelia. *Brain Spine* 2025 Jul 24;5:104336

Publications 2024

Chiu C, Kuchler A, Depienne C, Preuße C, Marina AD, Reis A, Kaiser FJ, **Nolte K**, Hentschel A, Schara-Schmidt U, Kölbel H, Roos A. Skeletal muscle vulnerability in a child with Pitt-Hopkins syndrome. *Skeletal Muscle* 2024 Jul 18;14(1): 15. doi: 10.1186/s13395-024-00348-0, PMID: 39026379

Gilliam E, **Achenbach P**, Suemmermann GJ, Wessely MN, Rossmanith P, Dohrn MF, Schulz JB, Waschbisch A, Brunkhorst R. Assessing hand motor function in chronic immune-mediated neuropathies: a proof-of-concept study using a data glove *J Neuroeng Rehabil*. 2024 Dec 20;21(1): 218. doi: 10.1186/s12984-024-01518-3. PMID: 39707391

Parlak A, Mueller CA, **Nolte KW**, Schmidt TP, Bertram U, Clusmann H, Blume C. Cervical myelopathy caused by IgG4-related hypertrophic spinal pachymeningitis: Case report and a descriptive review of the literature. *Brain Spine*. 2024 Sep 10;4:103325. doi: 10.1016/j.bas.2024.103325. eCollection 2024. PMID: 39558920

Schäfer B, Freund G, Orr J, **Nolte K**, **Weis J**, Bahm J, Beier JP. Technique and expected benefit of intraoperative perfusion imaging of peripheral nerves. *Plast Reconstr Surg Glob Open*. 2024 Nov 5;12(11): e6281. doi: 10.1097/GOX.0000000000006281. PMID: 39507316; PMCID: PMC11537564.

Mensch A, Jordan B, **Weis J**, **Nikolin S**, Schneider I, Abicht A, Gehling S, Kendzierski T, Stoltenburg-Didinger G, Stoevesandt D, Kraya T, Zierz S, Naegel S. A novel MYH14 variant presenting as a new phenotype of MYH14-associated neuromuscular disorders-Clinicohistologic findings and review of the literature. *J Clin Neuromuscul Dis*. 26(2):55-62, 2024

Kraft F, Rodriguez-Aliaga P, Yuan W, Franken L, **Zajt K**, Hasan D, Lee TT, Flex E, Hentschel A, Innes AM, Zheng B, Julia Suh DS, Knopp C, Lausberg E, Krause J, **Zhang X**, Trapane P, Carroll R, McClatchey M, Fry AE, Wang L, Giesselmann S, Hoang H, Baldrige D, Silverman GA, Radio FC, Bertini E, Cioffi A, Blood KA, de Sainte Agathe JM, Charles P, Bergant G, Čuturilo G, Peterlin B, Diderich K, Streff H, Robak L, Oegema R, van Binsbergen E, Herriges J, Saunders CJ, Maier A, Wolking S, Weber Y, Lochmüller H, Meyer S, Aleman A, Polavarapu K, Nicolas G, Goldenberg A, Guyant L, Pope K, Hehmeyer KN, Monaghan KG, Quade A, Smol T, Caumes R, Duerinckx S, Depondt C, Van Paesschen W, Rieubland C, Poloni C, Guipponi M, Arcioni S, Meuwissen M, Jansen AC, Rosenblum J, Haack TB, Bertrand M, Gerstner L, Magg J, Riess O, Schulz JB, Wagner N, Wiesmann M, **Weis J**, Eggermann T, Begemann M, Roos A, Häusler M, Schedl T, Tartaglia M, **Bremer J**, Pak SC, Frydman J, Elbracht M, Kurth I. Brain malformations and seizures by impaired chaperonin function of TRiC. *Science*. 2024 Nov; 386 (6721): 516-525. doi: 10.1126/science.adp8721. PMID:39480921

Holzer MT, Uruha A, Roos A, Hentschel A, Schänzer A, **Weis J**, Claeys KG, Schoser B, Montagnese F, Goebel HH, Huber M, Léonard-Louis S, Kötter I, Streichenberger N, Gallay L, Benveniste O, Schneider U, Preusse C, Krusche M, Stenzel W. Anti-Ku + myositis: an acquired inflammatory protein-aggregate myopathy. *Acta Neuropathol*. 2024 Jul 16;148(1): 6. doi: 10.1007/s00401-024-02765-3. PMID: 39012547; PMCID: PMC11252205.

Godbole S, Voß H, Gocke A, Schlumbohm S, Schumann Y, Peng B, Mynarek M, Rutkowski S, Dottermusch M, Dorostkar MM, Korshunov A, Mair T, Pfister SM, Kwiatkowski M, Hotze M, Neumann P, Hartmann C, **Weis J**, Liesche-Starnecker F, Guan Y, Moritz M, Siebels B, Struve N, Schlüter H, Schüller U, Krisp C, Neumann JE. Multiomic profiling of medulloblastoma reveals subtype-specific targetable

alterations at the proteome and N-glycan level. *Nat Commun.* 2024 Jul 24;15(1): 6237. doi: 10.1038/s41467-024-50554-z. PMID: 39043693; PMCID: PMC11266559.

Roos A, Häusler M, Kollipara L, Topf A, Preusse C, Stucka R, **Nolte K**, Strom T, Berutti R, Jiang X, Koll R, Lochmüller H, Schacht SM, Zahedi RP, **Weis J**, Senderek J. HNRNPA1 de novo Variant Associated with Early Childhood Onset, Rapidly Progressive Generalized Myopathy. *J Neuromuscul Dis.* 2024;11(5): 1131-1137. doi: 10.3233/JND-240050. PMID: 39121134; PMCID: PMC11380306.

Becker MHJ, Lassner F, **Nolte KW**, **Brook GA**, **Weis J**. The role of length of nerve grafts in combination with free functional muscle transplantation for brachial plexus injury: A single-center experience. *J Pers Med.* 2024 Sep 4;14(9): 940. doi: 10.3390/jpm14090940. PMID: 39338194; PMCID: PMC11433337.

Haag N*, **Bremer J***, Zempel* H. Understanding genetics, sex and signaling: Implications of sex-dependent APOE4-neutrophil-microglia interactions for Alzheimer's and tauopathies. *Signal Transduct Target Ther.* 2024 Sep 23;9(1): 252. doi: 10.1038/s41392-024-01967-1. PMID: 39313493. *contributed equally

Caredio D, Koderman M, Frontzek KJ, Sorce S, Nuvolone M, **Bremer J**, Mariutti G, Schwarz P, Madrigal L, Mitrovic M, Sellitto S, Streichenberger N, Scheckel C, Aguzzi A (2024): Prion diseases disrupt glutamate/glutamine metabolism in skeletal muscle. *PLoS Pathog.* 20 (9): e1012552. doi: 10.1371/journal.ppat.1012552. PMID: 39259763

Grübel N, Antoniadis G, Uerschels AK, Gembruch O, Marschal V, Deininger S, König R, Pala A, **Bremer J**, Dengler NF, Reuter M, Wirtz CR, Pedro MT (2024) : Collection of Rare Peripheral Nerve Tumors-Insights from the German Registry. *Cancers (Basel).* 16 (14): 2599. doi: 10.3390/cancers16142599. PMID: 39061237

Redlich JP, Feuerhake F, **Weis J**, Schaadt N, Teuber-Hanselmann S, Buck C, Luttmann S, Eberle A, **Nikolin S**, Appenzeller A, Portmann A, Homeyer A. Applications of artificial intelligence in the analysis of histopathology images of gliomas: a review. *npj Imaging* 2, 16 (2024). <https://doi.org/10.1038/s44303-024-00020-8>

Grübel N, Antoniadis G, König R, Wirtz CR, **Bremer J**, Pala A, Reuter M, Pedro MT. Case report: Atypical neurofibromatous neoplasm with uncertain biological potential of the sciatic nerve and a wide-spread arteriovenous fistula mimicking a malignant peripheral nerve tumor in a young patient with neurofibromatosis type 1. *Front Oncol.* 2024 May 10;14:1391456. doi: 10.3389/fonc.2024.1391456. eCollection 2024. PMID: 38800392 Free PMC article.

Bremer J, **Meinhardt A**, **Katona I**, Senderek J, Kämmerer-Gassler EK, Roos A, Ferbert A, **Schröder JM**, **Nikolin S**, **Nolte K**, **Sellhaus B**, **Popzhelyazkova K**, Tacke F, Schara-Schmidt U, Neuen-Jacob E, de Groote CC, de Jonghe P, Timmerman V, Baets J, **Weis J**. Myelin protein zero mutation-related hereditary neuropathies: Neuropathological insight from a new nerve biopsy cohort. *Brain Pathol.* 2024 Jan;34(1):e13200. doi: 10.1111/bpa.13200.

Distelmaier F, Sezer A, Helm C, Waldmüller S, Seibt A, Gangfuß A, Kölbel H, Schara-Schmidt U, Yuksel D, Talim B, Mayatepek E, **Nikolin S**, **Weis J**, Roos A, Haack TB. Biallelic truncating variants in PACSIN3 cause childhood-onset myopathy with hyperCKaemia. *Brain.* 147(7): e45-e49, 2024

Della Marina A, Hentschel A, Czech A, Schara-Schmidt U, Preusse C, Laner A, Abicht A, Ruck T, **Weis J**, Choueiri C, Lochmüller H, Kölbel H, Roos A. Novel Genetic and Biochemical Insights into the Spectrum of NEFL-Associated Phenotypes. *J Neuromuscul Dis.* 2024; 11(3): 625-645. doi: 10.3233/JND-230230. PMID: 38578900.

Publications 2023

Trofimov A, Pavlov D, **Goswami A**, Gorlova A, Chaprov K, Umriukhin A, Kalueff A, Deykin A, Lesch KP, Anthony DC, Strekalova T. Lipopolysaccharide triggers exacerbated microglial activation, excessive cytokine release and behavioural disturbances in mice with truncated Fused-in-Sarcoma Protein (FUS). *Brain Behav Immun Health*. 33: 100686, 2023

Achenbach P, Hillerbrand L, Gerardo-Nava JL, Dievernich A, **Hodde D**, Sechi AS, Dalton PD, Pich A, **Weis J**, **Altinova H**, **Brook GA**. Function Follows Form: Oriented Substrate Nanotopography Overrides Neurite-Repulsive Schwann Cell-Astrocyte Barrier Formation in an *In Vitro* Model of Glial Scarring. *Nano Lett*. 23(14): 6337-6346, 2023

Antoniani F, Cimino M, Mediani L, Vinet J, Verde EM, Secco V, **Yamoah A**, **Tripathi P**, Aronica E, Cicardi ME, Trotti D, Sternecker J, **Goswami A**, Carra S. Loss of PML nuclear bodies in familial amyotrophic lateral sclerosis-frontotemporal dementia. *Cell Death Discov*. 9(1):248, 2023

Motaln H, Čerček U, **Yamoah A**, **Tripathi P**, Aronica E, **Goswami A**, Rogelj B. Abl kinase-mediated FUS Tyr526 phosphorylation alters nucleocytoplasmic FUS localization in FTLD-FUS. *Brain*. 2023 Oct 3;146(10):4088-4104.

Foronda H, Fu Y, Covarrubias-Pinto A, Bocker HT, González A, Seemann E, Franzka P, Bock A, Bhaskara RM, Liebmann L, Hoffmann ME, **Katona I**, Koch N, **Weis J**, Kurth I, Gleeson JG, Reggiori F, Hummer G, Kessels MM, Qualmann B, Mari M, Dikić I, Hübner CA. Heteromeric clusters of ubiquitinated ER-shaping proteins drive ER-phagy. *Nature*. 618(7964): 402-410, 2023

Roos A, van der Ven PFM, Alrohaif H, Kölbel H, Heil L, Della Marina A, **Weis J**, Aßent M, Beck-Wödl S, Barresi R, Töpf A, O'Connor K, Sickmann A, Kohlschmidt N, El Gizouli M, Meyer N, Daya N, Grande V, Bois K, Kaiser FJ, Vorgerd M, Schröder C, Schara-Schmidt U, Gangfuss A, Evangelista T, Röbisch L, Hentschel A, Grüneboom A, Fuerst DO, Kuechler A, Tzschach A, Depienne C, Lochmüller H. Bi-allelic variants of FILIP1 cause congenital myopathy, dysmorphism and neurological defects. *Brain*. 3;146(10):4200-4216, 2023

Bremer J, Friemann J, von Stillfried S, Boor P, **Weis J**. Reduced T-cell densities in cranial nerves of patients who died with SARS-CoV-2 infection. *Acta Neuropathologica Communications* 11(1): 10, 2023

Igharo D, Thiel JC, Rolke R, Akkaya M, **Weis J**, **Katona I**, Schulz JB, Maier A. Skin biopsy reveals generalized small fibre neuropathy in hypermobile Ehlers-Danlos syndromes. *Eur J Neurol*. 30(3): 719-728, 2023

Yamoah A, **Tripathi P**, **Guo H**, **Scheve L**, Walter P, Johnen S, Müller F, **Weis J**, **Goswami A**. Early Alterations of RNA Binding Protein (RBP) Homeostasis and ER Stress-Mediated Autophagy Contributes to Progressive Retinal Degeneration in the *rd10* Mouse Model of Retinitis Pigmentosa (RP). *Cells*. 2023 Apr 6;12(7):1094. doi: 10.3390/cells12071094. PMID: 37048167; PMCID: PMC10092976.

Franzka P, Schüler SC, Kentache T, Storm R, Bock A, **Katona I**, **Weis J**, Buder K, Kaether C, Hübner CA. Impact of Hypermannosylation on the Structure and Functionality of the ER and the Golgi Complex. *Biomedicines*. 2023 Jan 6;11(1):146. doi: 10.3390/biomedicines11010146. PMID: 36672654; PMCID: PMC9856158.

Schänzer A, Dittmayer C, Porubsky S, **Weis J**, Goebel HH, Stenzel W. Neuropathologie I: Muskuläre Erkrankungen [Neuropathology I: muscular diseases]. *Pathologie* (Heidelb). 44(2): 104-112, 2023

Schänzer A, Dittmayer C, **Weis J**, Stenzel W, Goebel HH. Neuropathologie II: Erkrankungen des zentralen und peripheren Nervensystems: Ausblick auf neue Techniken in der Elektronenmikroskopie [Neuropathology II: diseases of the central and peripheral nervous systems: Outlook on new techniques in electron microscopy]. *Pathologie* (Heidelb). 44(2): 113-120, 2023

Publications 2022

Jackson J, Wischhof L, Scifo E, Pellizzer A, Wang Y, Piazzesi A, Gentile D, Siddig S, Stork M, Hopkins CE, Händler K, **Weis J**, Roos A, Schultze JL, Nicotera P, Ehninger D, Bano D. SGPL1 stimulates VPS39 recruitment to the mitochondria in MICU1 deficient cells. *Mol Metab*. 61: 101503, 2022

Jonigk D, Werlein C, Acker T, Aepfelbacher M, Amann KU, Baretton G, Barth P, Bohle RM, Büttner A, Büttner R, Dettmeyer R, Eichhorn P, Elezkurtaj S, Esposito I, Evert K, Evert M, Fend F, Gaßler N, Gattenlöhner S, Glatzel M, Göbel H, Gradhand E, Hansen T, Hartmann A, Heinemann A, Heppner FL, Hilsenbeck J, Horst D, Kamp JC, Mall G, Märkl B, Ondruschka B, Pablik J, Pfefferle S, Quaas A, Radbruch H, Röcken C, Rosenwald A, Roth W, Rudelius M, Schirmacher P, Slotta-Huspenina J, Smith K, Sommer L, Stock K, Ströbel P, Strobl S, Titze U, Weirich G, **Weis J**, Werner M, Wickenhauser C, Wiech T, Wild P, Welte T, von Stillfried S, Boor P. Organ manifestations of COVID-19: what have we learned so far (not only) from autopsies? *Virchows Arch*. 481(2): 139-159, 2022

Yilmazer-Hanke D, Ouali Alami N, Fang L, Klotz S, Kovacs GG, Pankratz H, **Weis J**, **Katona I**, Scheuerle A, Streit WJ, Del Tredici K. Differential Glial Chitotriosidase 1 and Chitinase 3-like Protein 1 Expression in the Human Primary Visual Cortex and Cerebellum after Global Hypoxia-Ischemia. *Neuroscience*. 506: 91-113, 2022

Hasselblatt M, Thomas C, Federico A, Nemes K, Johann PD, Bison B, Bens S, Dahlum S, Kordes U, Redlich A, Lessel L, Pajtler KW, Mawrin C, Schüller U, **Nolte K**, Kramm CM, Hinz F, Sahm F, Giannini C, Penkert J, Kratz CP, Pfister SM, Siebert R, Paulus W, Kool M, Frühwald MC. SMARCB1-deficient and SMARCA4-deficient Malignant Brain Tumors With Complex Copy Number Alterations and TP53 Mutations May Represent the First Clinical Manifestation of Li-Fraumeni Syndrome. *Am J Surg Pathol*. 46(9): 1277-1283, 2022

Clusmann J, Franco KC, Suárez DAC, **Katona I**, Minguez MG, Boersch N, Pissas KP, Vanek J, Tian Y, Gründer S. Acidosis induces RIPK1-dependent death of glioblastoma stem cells via acid-sensing ion channel 1a. *Cell Death Dis*. 13(8): 702, 2022

Comini M, Sierra-Marquez J, Guzman G, Franzen A, Willuweit A, **Katona I**, Hidalgo P, Fahlke C, Guzman RE. CLC Anion/Proton Exchangers Regulate Secretory Vesicle Filling and Granule Exocytosis in Chromaffin Cells. *J Neurosci*. 42(15): 3080-3095, 2022

Dohrn MF, Dumke C, Hornemann T, **Nikolin S**, Lampert A, Espenkott V, Vollert J, Ouwenbroek A, Zanella M, Schulz JB, Gess B, Rolke R. Deoxy-sphingolipids, oxidative stress, and vitamin C correlate with qualitative and quantitative patterns of small fiber dysfunction and degeneration. *Pain*. 163(9): 1800-1811, 2022

von Stillfried S, Bülow RD, Röhrig R, Boor P; German Registry of COVID-19 Autopsies (DeRegCOVID), DeRegCOVID Collaborators. First report from the German COVID-19 autopsy registry. *Lancet Reg Health Eur*. 2022 Feb 18; 15: 100330. doi: 10.1016/j.lanepe.2022.100330. eCollection 2022 Apr. PMID: 35531493

von Stillfried S, Bülow RD, Röhrig R, Meybohm P, Boor P; German Registry of COVID-19 Autopsies (DeRegCOVID), DeRegCOVID Collaborators#. Intracranial hemorrhage in COVID-19 patients during

extracorporeal membrane oxygenation for acute respiratory failure: a nationwide register study report. *Crit Care*. 2022 Mar 28; 26(1): 83. doi: 10.1186/s13054-022-03945-x. PMID: 35346314

Pilotto F, Schmitz A, Maharjan N, Diab R, Odriozola A, **Tripathi P**, **Yamoah A**, Scheidegger O, Oestmann A, Dennys CN, Sinha Ray S, Rodrigo R, Kolb S, Aronica E, Di Santo S, Widmer HR, Charlet-Berguerand N, Selvaraj BT, Chandran S, Meyer K, Zuber B, **Goswami A**, **Weis J**, Saxena S. PolyGA targets the ER stress-adaptive response by impairing GRP75 function at the MAM in C9ORF72-ALS/FTD. *Acta Neuropathol* 144(5):939-966, 2022

Maier A, Kapfenberger R, **Katona I**, **Weis J**, Schulz JB, Rolke R. Nonregional small fibre neuropathy in cases of autoimmune autonomic neuropathy. *J Neurol*. 269(12): 6648-6654, 2022

Altinova H, **Achenbach P**, **Palm M**, **Katona I**, Hermans E, Clusmann H, **Weis J**, **Brook GA**. Characterization of a Novel Aspect of Tissue Scarring Following Experimental Spinal Cord Injury and the Implantation of Bioengineered Type-I Collagen Scaffolds in the Adult Rat: Involvement of Perineurial-Like Cells? *Int. J. Mol. Sci.* 23(6), 3221, 2022

Božič J, Motaln H, Janež AP, Markič L, **Tripathi P**, **Yamoah A**, Aronica E, Lee YB, Heilig R, Fischer R, Thompson AJ, **Goswami A**, Rogelj B. Interactome screening of C9orf72 dipeptide repeats reveals VCP sequestration and functional impairment by polyGA. *Brain*. 145(2). 684-699, 2022

Dohrn MF, Heller C, Zengeler D, Obermaier CD, Biskup S, **Weis J**, **Nikolin S**, Claeys KG, Schöne U, Beijer D, Winter N, **Achenbach P**, Gess B, Schulz JB, Mulahasanovic L. Heterozygous POLG variant Ser1181Asn co-segregating in a family with autosomal dominant axonal neuropathy, proximal muscle fatigability, ptosis, and ragged red fibers. *Neurol Res Pract*. 4(1): 5, 2022

Dievernich A, **Achenbach P**, Davies L, Klinge U. Characterization of innate and adaptive immune cells involved in the foreign body reaction to polypropylene meshes in the human abdomen. *Hernia*. 26(1): 309-323, 2022

Liu J, Veldeman M, Höllig A, **Nolte K**, Liebenstund L, Willuweit A, Langen KJ, Rossaint R, Coburn M. Post-stroke treatment with argon preserved neurons and attenuated microglia/macrophage activation long-termly in a rat model of transient middle cerebral artery occlusion (tMCAO). *Sci Rep* 12(1): 691, 2022

Koeppen S, Hense J, **Nolte KW**, **Weis J**. Immune-mediated neuropathy related to bortezomib in a patient with multiple myeloma. *Arch Pathol Clin Res*. 6: 001-004, 2022

Dalton PD, O'Neill KL, Pêgo AP, Plant GW, Nisbet DR, Oudega M, **Brook GA**, Harvey AR (2022) Tissue Engineering of the Nervous System. In: *Tissue Engineering* 3rd Edition. Chapter 20. Clemens Van Blitterswijk, Jan De Boer (eds.). Academic Press - Elsevier. ISBN: 9780128244593

Weis J, **Katona I** (2022). Histopathologie der Haut- und Darminnervation. Kapitel 3, pp. 68-76, in C.-A. Haensch (Hrsg.) „Das autonome Nervensystem. Grundlagen, Organsysteme und Krankheitsbilder“. Verlag W. Kohlhammer, Stuttgart

Publications 2021

Ferreira N, Richner M, van der Laan A, Bergholdt Jul Christiansen I, Vægter CB, Nyengaard JR, Halliday GM, **Weis J**, Giasson BI, Mackenzie IR, Jensen PH, Jan A. Prodromal neuroinvasion of pathological α -synuclein in brainstem reticular nuclei and white matter lesions in a model of α -synucleinopathy. *Brain Commun*. 3(2): fcab104, 2021

Della Marina A, Arlt A, Schara-Schmidt U, Depienne C, Gangfuß A, Kölbel H, Sickmann A, Freier E, Kohlschmidt N, Hentschel A, **Weis J**, Czech A, Grüneboom A, Roos A. Phenotypical and Myopathological Consequences of Compound Heterozygous Missense and Nonsense Variants in SLC18A3. *Cells*. 10(12):3481, 2021

Weis J, Katona I, Nikolin S, Nobbio L, Prada V, Grandis M, Schenone A. Techniques for the standard histological and ultrastructural assessment of nerve biopsies. *J Peripher Nerv Syst*. Suppl 2:S3-S10, 2021

Achenbach P, Hambeukers I, Pierling AL, Gerardo-Nava JL, Hillerbrand L, Sechi AS, **Glücks KJ**, Dalton PD, Pich A, Dievernich A, **Altinova H, Brook GA**. A novel in vitro assay for peripheral nerve-related cell migration that preserves both extracellular matrix-derived molecular cues and nanofiber-derived topography. *J Neurosci Methods*. 361: 109289, 2021

Korinthenberg R, Trollmann R, Plecko B, Stettner GM, Blankenburg M, **Weis J**, Schoser B, Müller-Felber W, Lochbuehler N, Hahn G, Rudnik-Schöneborn S. Differential Diagnosis of Acquired and Hereditary Neuropathies in Children and Adolescents-Consensus-Based Practice Guidelines. *Children (Basel)*. 8(8):687, 2021 Deutsche Fassung: Differentialdiagnose der erworbenen und hereditären Neuropathien im Kindes- und Jugendalter. In: *Leitlinien Kinder- und Jugendmedizin*, Dt. Ges. für Kinder- und Jugendmedizin. Hrsg.: Niehues, T et al. Q9, 1-23. Elsevier, Urban & Fischer Verlag 2021

Mediani L, Antoniani F, Galli V, Vinet J, Carrà AD, Bigi I, Tripathy V, Tiago T, Cimino M, Leo G, Amen T, Kaganovich D, Cereda C, Pansarasa O, Mandrioli J, **Tripathi P**, Troost D, Aronica E, Buchner J, **Goswami A**, Sternecker J, Alberti S, Carra S. Hsp90-mediated regulation of DYRK3 couples stress granule disassembly and growth via mTORC1 signaling. *EMBO Rep*. 22(5): e51740, 2021

Freischmidt A, **Goswami A**, Limm K, Zimyanin VL, Demestre M, Glaß H, Holzmann K, Helferich AM, Brockmann SJ, **Tripathi P, Yamoah A**, Poser I, Oefner PJ, Böckers TM, Aronica E, Ludolph AC, Andersen PM, Hermann A, **Weis J**, Reinders J, Danzer KM, Weishaupt JH. A serum microRNA sequence reveals fragile X protein pathology in amyotrophic lateral sclerosis. *Brain*. 144(4): 1214-1229, 2021

Tripathi P, Guo H, Dreser A, Yamoah A, Sechi A, **Jesse C, Katona I**, Doukas P, **Nikolin S**, Ernst S, Aronica E, Troost D, Glass H, Hermann A, Steinbusch H, Feller A, Bergmann M, Jaarsma D*, **Weis J***, **Goswami A***. Pathomechanisms of ALS8: Altered autophagy and defective RNA binding protein (RBP) homeostasis due to the VAPB-P56S mutation. *Cell Death Dis*. 12(5): 466, 2021. *equal contribution

Lausberg E, Gießelmann S, Dewulf JP, Wiame E, Holz A, Salvarinova R, Van Karnebeek C, Klemm P, Ohl K, Mull M, Braunschweig T, **Weis J**, Sommer C, Demuth S, Haase C, Debray F-G, Libioulle C, Choukair D, Oommen PT, Borkhardt A, Surowy H, Wiczorek D, Meyer R, Eggermann T, Begemann M, Van Schaftingen E, Häusler M, Tenbrock K, van den Heuvel L, Elbracht M, Kurth, Kraft F. A human multisystem disorder with autoinflammation, leukoencephalopathy and hepatopathy is caused by mutations in C2orf69. *J Clin Invest*. 131(12): e143078, 2021

Peters S, Kuespert S, Wirkert E, Heydn R, Jurek B, Johannesen S, Hsam O, Korte S, Ludwig FT, Mecklenburg L, Mrowetz H, Altendorfer B, Poupardin R, Petri S, Thal DR, Hermann A, Weishaupt JH, **Weis J**, Aksoylu IS, Lewandowski SA, Aigner L, Bruun TH, Bogdahn U. Reconditioning the Neurogenic Niche of Adult Non-human Primates by Antisense Oligonucleotide-Mediated Attenuation of TGFβ Signaling. *Neurotherapeutics*. 18(3):1963-1979, 2021

Hrynevich A, **Achenbach P**, Jungst T, **Brook GA***, Dalton PD*. Design of Suspended Melt Electrowritten Fiber Arrays for Schwann Cell Migration and Neurite Outgrowth. *Macromol. Biosci*. 21(7): e2000439, 2021

Franzka P, Henze H, Jung MJ, Schüler SC, Mittag S, Biskup K, Liebmann L, Kentache T, Morales J, Martínez B, **Katona I**, Herrmann T, Huebner AK, Hennings JC, Groth S, Gresing LJ, Horstkorte R, Marquardt T, **Weis J**, Kaether C, Mutchinick OM, Ori A, Huber O, Blanchard V, von Maltzahn J, Hübner CA. GMPPA defects cause a neuromuscular disorder with α -dystroglycan hyperglycosylation. *J Clin Invest*. 131(9): e139076, 2021

Hummel C, Leylamian O, Pösch A, **Weis J**, Aronica E, Beyer C, Johann S. Expression and cell type-specific localization of inflammasome sensors in the spinal cord of SOD1^(G93A) mice and sALS patients. *Neuroscience*. 463: 288-302, 2021

Andereggen L, Mariani L, Beck J, Andres RH, Gralla J, Luedi MM, **Weis J**, Christ E. Lateral one-third gland resection in Cushing patients with failed adenoma identification leads to low remission rates: long-term observations from a small, single-center cohort. *Acta Neurochir (Wien)*. 163(11): 3161-3169, 2021

Deschauer M, Hengel H, Rupprich K, Kreiß M, Schlotter-Weigel B, Grimm M, Admard J, Schneider I, Alhaddad B, Gazou A, Sturm M, Vorgerd M, Balousha G, Balousha O, Falna M, Kirschke JS, Kornblum C, Jordan B, Kraya T, Strom TM, **Weis J**, Schöls L, Schara U, Zierz S, Riess O, Meitinger T, Haack TB. Bi-allelic truncating mutations in VWA1 cause neuromyopathy. *Brain*. 144(2): 574-583, 2021

Krasselt M, Schober R, **Weis J**, Baum P, Baerwald CGO, Seifert O. A Primary Myopathy Complicating Long-lasting Polymyalgia Rheumatica. *J Clin Rheumatol*. 27(1): e28-e29, 2021

Kohlschmidt N, Elbracht M, Czech A, Häusler M, Phan V, Töpf A, Huang KT, Bartok A, Eggermann K, Zippel S, Eggermann T, Freier E, Groß C, Lochmüller H, Horvath R, Hajnóczky G, **Weis J**, Roos A. Molecular pathophysiology of human MICU1-deficiency. *Neuropathol Appl Neurobiol*. 47(6):840-855, 2021

Ritschel N, Radbruch H, Herden C, Schneider N, Dittmayer C, Franz J, Thomas C, Silva Boos G, Pagenstecher A, Schulz-Schaeffer W, Stadelmann C, Glatzel M, Heppner FL, **Weis J**, Sohrabi K, Schänzer A, Németh A, Acker T; DGNN-Taskforce „CNS-COVID19“; „DEFEAT PANDEMIcs – Neuro-pathologische Referenzdiagnostik bei COVID-19“. COVID-19: Auswirkungen auf das zentrale und periphere Nervensystem [COVID-19 and the central and peripheral nervous system]. *Pathologe*. 42(2):172-182, 2021

Mihaylova V, Chablais F, **Bremer J**, Guggenberger R, Rushing EJ, Bethge T, Spiegel R, Jung HH. Collagen VI-Related Myopathy Caused by Compound Heterozygous Mutations of COL6A3 in a Consanguineous Kurdish Family. *J Clin Neuromuscul Dis*. 22(3): 173-179, 2021

Schröder JM, Zypen vd E. (2021) Normale Anatomie und Histologie des peripheren Nervensystems. In: Läsionen peripherer Nerven und radikuläre Syndrome. Kapitel 1.3, pp. 23-29. Müller-Vahl u. a. , Georg Thieme Verlag KG Stuttgart - New York

Schröder JM. (2021) Histopathologie der Läsionen und Regenerationsvorgänge im peripheren Nervensystem. In: Läsionen peripherer Nerven und radikuläre Syndrome. Kapitel 1.4, pp. 30. Müller-Vahl u. a. , Georg Thieme Verlag KG Stuttgart - New York

Schröder JM. (2021) Tumoren peripherer Nerven. In: Läsionen peripherer Nerven und radikuläre Syndrome. Kapitel 1.6, pp. 44-46. Müller-Vahl u. a. , Georg Thieme Verlag KG Stuttgart - New York

Publications 2020

Blume C, Tuleta I, **Nolte K**, Eichhorn KW, Jakob M, Clusmann H, Send T. Neurosarcoidosis As a Rare Differential Diagnosis for Single Or Multiple Lesions of the Nervous System. *Br J Neurosurg.* 34(5): 495-499, 2020

Kölbel H, Roos A, van der Ven PFM, Evangelista T, **Nolte K**, Johnson K, Töpf A, Wilson M, Kress W, Sickmann A, Straub V, Kollipara L, **Weis J**, Fürst DO, Schara U. First clinical and myopathological description of a myofibrillar myopathy with congenital onset and homozygous mutation in FLNC. *Hum Mutat.* 41(9): 1600-1614, 2020

Kremer B, Coburn M, Weinandy A, **Nolte K**, Clusmann H, Veldeman M, Höllig A. Argon treatment after experimental subarachnoid hemorrhage: evaluation of microglial activation and neuronal survival as a subanalysis of a randomized controlled animal trial. *Med Gas Res.* 10(3): 103-109, 2020

Rabenstein M, **Weis J**, Abicht A, Fink GR, Lehmann HC, Wunderlich G. [Multiple acyl-CoA dehydrogenase deficiency/glutaric aciduria type 2: difficult diagnosis, easy to treat]. *Nervenarzt.* 91(4): 349-352, 2020

Billig SCI, Schauermann JC, Rolke R, **Katona I**, Schulz JB, Maier A. Quantitative sensory testing predicts histological small fiber neuropathy in postural tachycardia syndrome. *Neurol Clin Pract.* 10(5): 428-434, 2020

Gerardo-Nava JL, Rose JC, **Altinova H**, Dalton PD, De Laporte L, **Brook GA** (2020) Nanofibres and Nanostructured Scaffolds for Nervous System Lesions. In: **Nanomedicines for Brain Drug Delivery** Chapter 3, pp. 61-101. Morales JO, Gaillard PJ (eds.) Nanomedicines for Brain Springer Protocols, Springer Science+Business Media, LLC, New York

Tian Y, Korn P, **Tripathi P**, Komnig D, Wiemuth D, Nikouee A, Classen A, Bolm C, Falkenburger BH, Lüscher B, Gründer S. The mono-ADP-ribosyltransferase ARTD10 regulates the voltage-gated K⁺ channel Kv1.1 through protein kinase C delta. *BMC Biol.* 18(1): 143, 2020

Heuschkel MA, Skenteris NT, Hutcheson JD, van der Valk DD, **Bremer J**, Goody P, Hjortnaes J, Jansen F, Bouten CVC, van den Bogaerd A, Matic L, Marx N, Goettsch C. Integrative Multi-Omics Analysis in Calcific Aortic Valve Disease Reveals a Link to the Formation of Amyloid-Like Deposits. *Cells.* 9(10): 2164, 2020

Doyen PJ, Beckers P, **Brook GA**, Hermans E. Regulators of G protein signalling as pharmacological targets for the treatment of neuropathic pain. *Pharmacol Res.* Aug 26; 160: 105148, 2020

Mair D, Biskup S, Kress W, Abicht A, Brück W, Zechel S, Knop KC, Koenig FB, **Tey S**, **Nikolin S**, Eggermann K, Kurth I, Ferbert A, **Weis J**. Differential diagnosis of vacuolar myopathies in the NGS era. *Brain Pathol.* 30(5): 877-896, 2020

Mathis S, Vallat JM, **Weis J**. When botany inspired pathology of the peripheral nervous system. *Neurology.* 95(12): 532-536, 2020

Hedberg-Oldfors C, Meyer R, **Nolte K**, Abdul Rahim Y, Lindberg C, Karason K, Thuestad IJ, Visuttijai K, Geijer M, Begemann M, Kraft F, Lausberg E, Hitpass L, Götzl R, Luna EJ, Lochmüller H, Koschmieder S, Gramlich M, Gess B, Elbracht M, **Weis J**, Kurth I, Oldfors A, Knopp C. Loss of supervillin causes myopathy with myofibrillar disorganization and autophagic vacuoles. *Brain.* 143(8): 2406-2420, 2020

Bremer J, Kottke R, Johann PD, von Hoff K, Brazzola P, Grotzer A, Kool M, Rushing E, Gerber NU. A single supratentorial high-grade neuroepithelial tumor with two distinct *BCOR* mutations, exceptionally

long complete remission and survival. *Pediatr Blood Cancer*. 67(7): e28384, 2020

Kulesa M, Weyer-Menkhoff I, Viergutz L, Kornblum C, Claeys KG, Schneider I, Plöckinger U, Young P, Boentert M, Vielhaber S, Mawrin C, Bergmann M, **Weis J**, Ziagaki A, Stenzel W, Deschauer M, Nolte D, Hahn A, Schoser B, Schänzer A. An integrative correlation of myopathology, phenotype, and genotype in late onset Pompe disease. *Neuropathol Appl Neurobiol*. 46(4): 359-374, 2020

Yamoah A, Tripathi P, Sechi A, Köhler C, **Guo H**, Chandrasekar A, **Nolte KW**, Wruck CJ, **Katona I**, Anink J, Troost D, Aronica E, Steinbusch H, **Weis J**, **Goswami A**. Aggregates of RNA Binding Proteins and ER Chaperones Linked to Exosomes in Granulovacuolar Degeneration of the Alzheimer's Disease Brain. *J Alzheimers Dis*. 75(1): 139-156, 2020

Yilmazer-Hanke D, Mayer T, Müller HP, Neugebauer H, Abaei A, Scheuerle A, **Weis J**, Forsberg KME, Althaus K, Meier J, Ludolph AC, Del Tredici K, Braak H, Kassubek J, Rasche V. Histological correlates of postmortem ultra-high-resolution single-section MRI in cortical cerebral microinfarcts. *Acta Neuropathol Commun*. 8(1): 33, 2020

Keijden H, Konrad J, Hoffmann B, Gerardo-Nava J, Rütten S, Merkel R, Vázquez-Jiménez J, **Brook GA**, Jockenhoevel S, Mela P. A bench-top molding method for the production of cell-laden fibrin microfibers with longitudinal topography. *J Biomed Mater Res B Appl Biomater*. 108(4): 1198-1212, 2020

Cadoni MPL, Biggio ML, Arru G, Secchi G, Orrù N, Clemente MG, Sechi G, **Yamoah A, Tripathi P**, Orrù S, Manetti R, Galleri G. VAPB ER-Aggregates, A Possible New Biomarker in ALS Pathology. *Cells*. 9(1): 164, 2020

Altinova H, Hammes S, Palm M, Achenbach P, Gerardo-Nava J, Deumens R, Führmann T, van Neerven SGA, Hermans E, **Weis J, Brook G**. Dense fibroadhesive scarring and poor blood vessel-maturation hamper the integration of implanted collagen scaffolds in an experimental model of spinal cord injury. *Biomed Mater*. 15(1): 015012, 2020

Farschtschi SC, Kluwe L, Schön G, Friedrich RE, Matschke J, Glatzel M, **Weis J**, Hagel C, Mautner VF. Distinctive low epidermal nerve fiber density in schwannomatosis patients provides a major parameter for diagnosis and differential diagnosis. *Brain Pathol*. 30(2): 386-391, 2020

Publications 2019

Dafsari HS, Kocaturk NM, Daimagüler HS, Brunn A, Dötsch J, **Weis J**, Deckert M, Cirak S. Bi-allelic mutations in uncoordinated mutant number-45 myosin chaperone B are a cause for congenital myopathy. *Acta Neuropathol Commun*. 7(1): 211, 2019

Rudnik-Schöneborn S, Huemer M, **Weis J**, Sauer E, Meng G. Early onset facioscapulohumeral muscular dystrophy - Long-term follow-up of a patient with total facial diplegia. *Neuromuscul Disord*. 29(12): 973-976, 2019

Brenner D, Rosenbohm A, Yilmaz R, Müller K, Grehl T, Petri S, Meyer T, Grosskreutz J, Weydt P, Ruf W, Neuwirth C, Weber M, Pinto S, Claeys KG, Schrank B, Jordan B, Knehr A, Günther K, Hübers A, Zeller D, Kubisch C, Jablonka S, Sendtner M, Klopstock T, de Carvalho M, Sperfeld A, Borck G, Volk AE, Dorst J, **Weis J**, Otto M, Schuster J, Del Tredici K, Braak H, Danzer KM, Freischmidt A, Meitinger T, Ludolph AC, Andersen PM, Weishaupt JH; German ALS network MND-NET. Reply: Adult-onset distal spinal muscular atrophy: a new phenotype associated with KIF5A mutations. *Brain*. 142(12): e67, 2019

Phan V, Cox D, Cipriani S, Spendiff S, Buchkremer S, O'Connor E, Horvath R, Goebel HH, Hathazi D, Lochmüller H, Straka T, Rudolf R, **Weis J**, Roos A. SIL1 deficiency causes degenerative changes of peripheral nerves and neuromuscular junctions in fish, mice and human. *Neurobiol Dis.* 124: 218-229, 2019

Prasad V, Wasser Y, Hans F, **Goswami A**, **Katona I**, Outeiro TF, Kahle PJ, Schulz JB, Voigt A. Monitoring α -synuclein multimerization in vivo. *FASEB J.* 33(2): 2116-2131, 2019

Stengel H, Vural A, Brunder AM, Heinius A, Appeltshauser L, Fiebig B, Giese F, Dresel C, Papagianni A, Birklein F, **Weis J**, Huchtemann T, Schmidt C, Körtvelyessy P, Villmann C, Meinl E, Sommer C, Leyboldt F, Doppler K. Anti-pan-neurofascin IgG3 as a marker of fulminant autoimmune neuropathy. *Neurol Neuroimmunol Neuroinflamm.* 6(5). pii: e603, 2019

Romeike BFM, Becker K, Großkreutz J, Schulz S, **Weis J**, Cirak S. A family with limb girdle muscular dystrophy type 1B and multiple exostoses. *Clin Neuropathol.* 38(5): 225-232, 2019

Liu J, **Nolte K**, **Brook G**, Liebenstund L, Weinandy A, Höllig A, Veldeman M, Willuweit A, Langen KJ, Rossaint R, Coburn M. Post-stroke treatment with argon attenuated brain injury, reduced brain inflammation and enhanced M2 microglia/macrophage polarization: a randomized controlled animal study. *Crit Care.* 23(1): 198, 2019

Munter JP, Beugels J, Munter S, Jansen L, Cillero-Pastor B, Moskvina O, **Brook G**, Pavlov D, Strekalova T, Kramer BW, E Ch W. Standardized human bone marrow-derived stem cells infusion improves survival and recovery in a rat model of spinal cord injury. *J Neurol Sci.* 402: 16-29, 2019

Gatz C, Hathazi D, Münchberg U, **Buchkremer S**, **Labisch T**, Munro B, Horvath R, Töpf A, **Weis J**, Roos A. Identification of Cellular Pathogenicity Markers for SIL1 Mutations Linked to Marinesco-Sjögren Syndrome. *Front Neurol.* 10: 562, 2019

Ross JA, Levy Y, Ripolone M, Kolb JS, Turmaine M, Holt M, Lindqvist J, Claeys KG, **Weis J**, Monforte M, Tasca G, Moggio M, Figeac N, Zammit PS, Jungbluth H, Fiorillo C, Vissing J, Witting N, Granzier H, Zanolini E, Hardeman EC, Wallgren-Pettersson C, Ochala J. Impairments in contractility and cytoskeletal organisation cause nuclear defects in nemaline myopathy. *Acta Neuropathol.* 138(3): 477-495, 2019

Joseph S, Vingill S, Jahn O, Fledrich R, Werner HB, **Katona I**, Möbius W, Mitkovski M, Huang Y, **Weis J**, Sereda MW, Schulz JB, Nave KA, Stegmüller J. Myelinating glia-specific deletion of Fbxo7 in mice triggers axonal degeneration in the central nervous system together with peripheral neuropathy. *J Neurosci.* 39(28): 5606-5626, 2019

Marrone L, Drexler HCA, Wang J, **Tripathi P**, Distler T, Heisterkamp P, Anderson EN, Kour S, Moraiti A, Maharana S, Bhatnagar R, Belgard TG, Tripathy V, Kalmbach N, Hosseinzadeh Z, Crippa V, Abo-Rady M, Wegner F, Poletti A, Troost D, Aronica E, Busskamp V, **Weis J**, Pandey UB, Hyman AA, Alberti S, **Goswami A**, Sternecker J. FUS pathology in ALS is linked to alterations in multiple ALS-associated proteins and rescued by drugs stimulating autophagy. *Acta Neuropathol.* 138(1): 67-84, 2019

Karsai G, Kraft F, Haag N, Korenke GC, Hänisch B, Othman A, Suriyanarayanan S, Steiner R, Knopp C, Mull M, Bergmann M, **Schröder JM**, **Weis J**, Elbracht M, Begemann M, Hornemann T, Kurth I. DEGS1-associated aberrant sphingolipid metabolism impairs nervous system function in humans. *J Clin Invest.* 129(3): 1229-1239, 2019

Fischbach F, Nedelcu J, Leopold P, Zhan J, Clarner T, Nellessen L, Beißel C, van Heuvel Y, **Goswami**

A, Weis J, Denecke B, Schmitz C, Hochstrasser T, Nyamoya S, Victor M, Beyer C, Kipp M. Cuprizone-induced graded oligodendrocyte vulnerability is regulated by the transcription factor DNA damage-inducible transcript 3. *Glia*. 67(2): 263-276, 2019

Kölbel H, Abicht A, Schwartz O, **Katona I**, Paulus W, Neuen-Jacob E, **Weis J**, Schara U. Characteristic clinical and ultrastructural findings in nesprinopathies. *Eur J Paediatr Neurol*. 23(2): 254-261, 2019

Altinova H*, **Hammes S***, **Palm M**, Gerardo-Nava J, **Achenbach P**, Deumens R, Hermans E, Führmann T, Boecker A, van Neerven S, Bozkurt A, **Weis J**, **Brook GA**. Fibroadhesive scarring of grafted collagen scaffolds interferes with implant–host neural tissue integration and bridging in experimental spinal cord injury. *Regenerative Biomaterials* 6(2): 75-87, 2019, *Equal contribution

Publications 2018

De Paepe B, Zschüntzsch J, Šokčević T, **Weis J**, Schmidt J, De Bleecker JL. Induction of Osmolyte Pathways in Skeletal Muscle Inflammation: Novel Biomarkers for Myositis. *Front Neurol*. 9: 846, 2018

Herbelet S, De Vlieghere E, Gonçalves A, De Paepe B, Schmidt K, Nys E, Weynants L, **Weis J**, Van Peer G, Vandesompele J, Schmidt J, De Wever O, De Bleecker JL. Localization and Expression of Nuclear Factor of Activated T-Cells 5 in Myoblasts Exposed to Pro-inflammatory Cytokines or Hyperosmolar Stress and in Biopsies from Myositis Patients. *Front Physiol*. 9: 126, 2018

Komnig D, Gertz K, Habib P, **Nolte KW**, Meyer T, Brockmann MA, Endres M, Rathkolb B, Hrabě de Angelis M; German Mouse Clinic Consortium, Schulz JB, Falkenburger BH, Reich A. Faim2 contributes to neuroprotection by erythropoietin in transient brain ischemia. *J Neurochem*. 145(3): 258-270, 2018

Brücken A, Bleilevens C, Berger P, **Nolte K**, Gaisa NT, Rossaint R, Marx G, Derwall M, Fries M. Effects of inhaled nitric oxide on outcome after prolonged cardiac arrest in mild therapeutic hypothermia treated rats. *Sci Rep*. 8(1): 6743, 2018

Forsberg KME, Zhang Y, Reiners J, Ander M, Niedermayer A, Fang L, Neugebauer H, Kassubek J, **Katona I**, **Weis J**, Ludolph AC, Del Tredici K, Braak H, Yilmazer-Hanke D. Endothelial damage, vascular bagging and remodeling of the microvascular bed in human microangiopathy with deep white matter lesions. *Acta Neuropathol Commun*. 6(1): 128, 2018

Vill K, Müller-Felber W, Gläser D, Kuhn M, Teusch V, Schreiber H, **Weis J**, Klepper J, Schirmacher A, Blaschek A, Wiessner M, Strom TM, Dräger B, Hofmeister-Kiltz K, Tacke M, Gerstl L, Young P, Horvath R, Senderek J. SACS variants are a relevant cause of autosomal recessive hereditary motor and sensory neuropathy. *Hum Genet*. 137(11-12): 911-919, 2018

Heinen MC, Babler A, **Weis J**, Elsas J, **Nolte K**, Kipp M, Jahnen-Dechent W, Häusler M. Fetuin-A protein distribution in mature inflamed and ischemic brain tissue. *PLoS One*. 13(11): e0206597, 2018

Quade A, **Weis J**, Kurth I, Rolke R, Bienert M, Schrading S, Rohrmann D, Yüksel Z, Häusler M. Microangiopathy and mild mixed neuromyopathic alterations in a patient with homozygous PIEZO-2 mutation. *Neuromuscul Disord*. 28(12): 1006-1011, 2018

Vettermann FJ, Felsberg J, Reifenberger G, Hasselblatt M, Forbrig R, Berding G, la Fougère C, Galldiks N, Schittenhelm J, **Weis J**, Albert NL, Schüller U. Characterization of Diffuse Gliomas With Histone H3-G34 Mutation by MRI and Dynamic 18F-FET PET. *Clin Nucl Med*. 43(12): 895-898, 2018

Kork F, Jankowski J, **Goswami A, Weis J, Brook G, Yamoah A**, Anink J, Aronica E, Fritz S, Huck C, Schipke C, Peters O, Tepel M, Noels H, Jankowski V. Golgin A4 in CSF and granulovacuolar degenerations of Alzheimer patients. *Neurology*. 91(19): e1799-e1808, 2018

González Coraspe JA, **Weis J**, Anderson ME, Münchberg U, Lorenz K, Buchkremer S, Carr S, Zahedi RP, **Brauers E**, Michels H, Sunada Y, Lochmüller H, Campbell KP, Freier E, Hathazi D, Roos A. Biochemical and pathological changes result from mutated Caveolin-3 in muscle. *Skelet Muscle*. 8(1): 28, 2018

Arndt P, Leistner ND, Neuss S, Kaltbeitzel D, **Brook GA**, Grosse J. Artificial urine and FBS supplemented media in cytocompatibility assays for PLGA-PEG-based intravesical devices using the urothelium cell line UROtsa. *J Biomed Mater Res B Appl Biomater*. 106(6): 2140-2147, 2018

Laššuthová P, Vill K, Erdem-Ozdamar S, **Schröder JM**, Topaloglu H, Horvath R, Müller-Felber W, Bansagi B, Schlotter-Weigel B, Gläser D, Neupauerová J, Sedláčková L, Staněk D, Mazanec R, **Weis J**, Seeman P, Senderek J. Novel SBF2 mutations and clinical spectrum of Charcot-Marie-Tooth neuropathy type 4B2. *Clin Genet*. 94(5): 467-472, 2018

Boecker AH, Bozkurt A, Kim BS, Altinova H, Tank J, Deumens R, Tolba R, **Weis J, Brook GA**, Pallua N, van Neerven SGA. Cell-enrichment with olfactory ensheathing cells has limited local extra beneficial effects on nerve regeneration supported by the nerve guide Perimaix. *J Tissue Eng Regen Med*. 12(11): 2125-2137, 2018

Wunderlich G, Brunn A, Daimagüler HS, Bozoglu T, Fink GR, Lehmann HC, **Weis J**, Cirak S. Long term history of a congenital core-rod myopathy with compound heterozygous mutations in the Nebulin gene. *Acta Myol*. 37(2): 121-127, 2018

Fledrich R, Abdelaal T, Rasch L, Bansal V, Schütza V, Brügger B, Luchtenborg C, Prukop T, Stenzel J, Rahman RU, Hermes D, Ewers D, Möbius W, Ruhwedel T, **Katona I, Weis J**, Klein D, Martini R, Brück W, Müller WC, Bonn S, Bechmann I, Nave KA, Stassart RM, Sereda MW. Targeting myelin lipid metabolism as a potential therapeutic strategy in a model of CMT1A neuropathy. *Nat Commun*. 9(1): 3025, 2018

Lohmann P, Piroth MD, **Sellhaus B, Weis J**, Geisler S, Oros-Peusquens AM, Mohlberg H, Amunts K, Shah NJ, Galldiks N, Langen KJ. Correlation of Dynamic O-(2-[18F]Fluoroethyl)-L-Tyrosine Positron Emission Tomography, Conventional Magnetic Resonance Imaging, and Whole-Brain Histopathology in a Pretreated Glioblastoma: A Postmortem Study. *World Neurosurg*. 119: e653-e660, 2018

Issop Y, Hathazi D, Khan MM, Rudolf R, **Weis J**, Spendiff S, Slater CR, Roos A, Lochmüller H. GFPT1 deficiency in muscle leads to myasthenia and myopathy in mice. *Hum Mol Genet*. 27(18): 3218-3232, 2018

Nikoubashman O, Heringer S, Feher K, Brockmann MA, **Sellhaus B, Dreser A**, Kurtenbach K, Pjontek R, Jockenhövel S, **Weis J**, Kießling F, Gries T, Wiesmann M. Development of a Polymer-Based Biodegradable Neurovascular Stent Prototype: A Preliminary In Vitro and In Vivo Study. *Macromol Biosci*. 18(7): e1700292, 2018

Yuan X, Klein D, Kerscher S, West BL, **Weis J, Katona I**, Martini R. Macrophage depletion ameliorates peripheral neuropathy in aging mice. *J Neurosci*. 38(19): 4610-4620, 2018

Paul NE, Denecke B, Kim BS, **Dreser A**, Bernhagen J, Pallua N. The effect of mechanical stress on the proliferation, adipogenic differentiation and gene expression of human adipose-derived stem cells. *J Tissue Eng Regen Med*. 12(1): 276-284, 2018

Dusanic M, Dekomien G, Lücke T, Vorgerd M, **Weis J**, Epplen JT, Köhler C, Hoffjan S. Novel Nonsense Mutation in SLC39A13 Initially Presenting as Myopathy: Case Report and Review of the Literature. *Mol Syndromol*. 9(2): 100-109, 2018

Müller K, Brenner D, Weydt P, Meyer T, Grehl T, Petri S, Grosskreutz J, Schuster J, Volk AE, Borck G, Kubisch C, Klopstock T, Zeller D, Jablonka S, Sendtner M, Klebe S, Knehr A, Günther K, **Weis J**, Claeys KG, Schrank B, Sperfeld AD, Hübers A, Otto M, Dorst J, Meitinger T, Strom TM, Andersen PM, Ludolph AC, Weishaupt JH; German ALS network MND-NET. Comprehensive analysis of the mutation spectrum in 301 German ALS families. *J Neurol Neurosurg Psychiatry*. 89(8): 817-827, 2018

Lehmann S, Esch E, Hartmann P, **Goswami A**, **Nikolin S**, **Weis J**, Beyer C, Johann S. Expression profile of pattern recognition receptors in skeletal muscle of SOD1((G93A)) amyotrophic lateral sclerosis (ALS) mice and sporadic ALS patients. *Neuropathol Appl Neurobiol*. 44(6): 606-627, 2018

Brenner D, Yilmaz R, Müller K, Grehl T, Petri S, Meyer T, Grosskreutz J, Weydt P, Ruf W, Neuwirth C, Weber M, Pinto S, Claeys KG, Schrank B, Jordan B, Knehr A, Günther K, Hübers A, Zeller D; German ALS network MND-NET, Kubisch C, Jablonka S, Sendtner M, Klopstock T, de Carvalho M, Sperfeld A, Borck G, Volk AE, Dorst J, **Weis J**, Otto M, Schuster J, Del Tredici K, Braak H, Danzer KM, Freischmidt A, Meitinger T, Strom TM, Ludolph AC, Andersen PM, Weishaupt JH. Hot-spot KIF5A mutations cause familial ALS. *Brain*. 141(3): 688-697, 2018

Naumann M, Pal A, **Goswami A**, Lojewski X, Japtok J, Vehlow A, Naujock M, Günther R, Jin M, Stanslowsky N, Reinhardt P, Sternecker J, Frickenhaus M, Pan-Montojo F, Storkebaum E, Poser I, Freischmidt A, Weishaupt JH, Holzmann K, Troost D, Ludolph AC, Boeckers TM, Liebau S, Petri S, Cordes N, Hyman AA, Wegner F, Grill SW, **Weis J**, Storch A, Hermann A. Impaired DNA damage response signaling by FUS-NLS mutations leads to neurodegeneration and FUS aggregate formation. *Nat Commun*. 9(1): 335, 2018

Bouhy D, Juneja M, **Katona I**, Holmgren A, Asselbergh B, De Winter V, Hochepped T, Goossens S, Haigh JJ, Libert C, Ceuterick-de Groote C, Irobi J, **Weis J**, Timmerman V. A knock-in/knock-out mouse model of HSPB8-associated distal hereditary motor neuropathy and myopathy reveals toxic gain-of-function of mutant Hspb8. *Acta Neuropathol*. 135(1): 131-148, 2018

Breuer T, Bleilevens C, Rossaint R, Marx G, Gehrenkemper J, Dierksen H, Delpierre A, **Weis J**, Gayan-Ramirez G, Bruells CS. Dexmedetomidine Impairs Diaphragm Function and Increases Oxidative Stress but Does Not Aggravate Diaphragmatic Atrophy in Mechanically Ventilated Rats. *Anesthesiology*. 128(4): 784-795, 2018

Eggermann K, Gess B, Häusler M, **Weis J**, Hahn A, Kurth I. Hereditary Neuropathies. *Dtsch Arztebl Int*. 115(6): 91-97, 2018

De Berdt P, Bottemanne P, Bianco J, Alhouayek M, Diogenes A, Llyod A, **Gerardo-Nava J**, **Brook GA**, Miron V, Muccioli GG, Rieux AD. Stem cells from human apical papilla decrease neuro-inflammation and stimulate oligodendrocyte progenitor differentiation via activin-A secretion. *Cell Mol Life Sci*. 75(15): 2843-2856, 2018

Labisch T, **Buchkremer S**, Phan V, Kollipara L, **Gatz C**, **Lentz C**, **Nolte K**, Vervoorts-Weber J, **González Coraspe JA**, Sickmann A, Carr S, Zahedi RP, **Weis J**, **Roos A**. Tracking effects of SIL1 increase: taking a closer look beyond the consequences of elevated expression level. *Molecular Neurobiology* 55(3): 2524-2546, 2018

Radke J, Koll R, Preuße C, Pehl D, Todorova K, Schönemann C, Allenbach Y, Aronica E, de Visser M, Heppner FL, **Weis J**, Doostkam S, Maissonobe T, Benveniste O, Goebel HH, Stenzel W. Architectural B-cell organization in skeletal muscle identifies subtypes of dermatomyositis. *Neurol Neuroimmunol Neuroinflamm.* 5(3): e451, 2018

Publications 2017

Wang H, Salter CG, Refai O, Hardy H, Barwick KES, Akpulat U, Kvarnung M, Chioza BA, Harlalka G, Taylan F, Sejersen T, Wright J, Zimmerman HH, Karakaya M, Stüve B, **Weis J**, Schara U, Russell MA, Abdul-Rahman OA, Chilton J, Blakely RD, Baple EL, Cirak S, Crosby AH. Choline transporter mutations in severe congenital myasthenic syndrome disrupt transporter localization. *Brain.* 140(11): 2838-2850, 2017

Katona I, Weis J. Diseases of the peripheral nerves. *Handb Clin Neurol.* 145: 453-474, 2017

Jesse CM, Bushuven E, Tripathi P, Chandrasekar A, Simon CM, Drepper C, **Yamoah A, Dreser A, Katona I,** Johann S, Beyer C, Wagner S, Grond M, **Nikolin S,** Anink J, Troost D, Sendtner M, **Goswami A*, Weis J*.** ALS-associated endoplasmic reticulum proteins in denervated skeletal muscle: Implications for motor neuron disease pathology. *Brain Pathol.* 27(6): 781-794, 2017

Kollipara L, Buchkremer S, Coraspe JAG, Hathazi D, Senderek J, **Weis J,** Zahedi RP, Roos A. In-depth phenotyping of lymphoblastoid cells suggests selective cellular vulnerability in Marinesco-Sjögren syndrome. *Oncotarget.* 8(40): 68493-68516, 2017

Bozkurt A, Claeys KG, Schrading S, Rödler JV, Altinova H, Schulz JB, **Weis J,** Pallua N, van Neerven SGA. Clinical and biometrical 12-month follow-up in patients after reconstruction of the sural nerve biopsy defect by the collagen-based nerve guide Neuromaix. *Eur J Med Res.* 22(1): 34, 2017

Quade A, Wiesmann M, **Weis J,** Kurth I, Jalaie H, Rohrbach M, Häusler M. Stroke in Ehlers-Danlos Syndrome Kyphoscoliotic Type: Dissection or Vasculitis? *Pediatr Neurol.* 74: 92-96, 2017

Dreser A, Vollrath JT, Sechi A, Johann S, **Roos A, Yamoah A, Katona I,** Boholega S, Wiemuth D, Tian Y, Schmidt A, Vervoorts-Weber J, Dohmen M, Beyer C, Anink J, Aronica E, Troost D, **Weis J*, Goswami A*.** The ALS-linked E102Q mutation in Sigma receptor-1 leads to ER stress-mediated defects in protein homeostasis and dysregulation of RNA binding proteins. *Cell Death Differ.* 24(10): 1655-1671, 2017 *equal contribution

Cordts I, Bodart N, Hartmann K, Karagiorgou K, Tzartos JS, Mei L, Reimann J, Van Damme P, Rivner MH, Vigneron A, **Weis J,** Schulz JB, Tzartos SJ, Claeys KG. Screening for lipoprotein receptor-related protein 4-, agrin-, and titin-antibodies and exploring the autoimmune spectrum in myasthenia gravis. *J Neurol.* 264(6): 1193-1203, 2017

Schnitzler LJ, Schreckenbach T, Nadaj-Pakleza A, Stenzel W, Rushing EJ, Van Damme P, Ferbert A, Petri S, Hartmann C, Bornemann A, Meisel A, Petersen JA, Tousseyn T, Thal DR, Reimann J, De Jonghe P, Martin JJ, Van den Bergh PY, Schulz JB, **Weis J,** Claeys KG. Sporadic late-onset nemaline myopathy: clinico-pathological characteristics and review of 76 cases. *Orphanet J Rare Dis.* 12(1): 86, 2017

Fragoulis A, Siegl S, Fendt M, Jansen S, Soppa U, Brandenburg LO, Pufe T, **Weis J,** Wruck CJ. Oral administration of methysticin improves cognitive deficits in a mouse model of Alzheimer's disease. *Redox Biol.* 12: 843-853, 2017

Weis J, Claeys KG, Roos A, **Azzedine H**, **Katona I**, **Schröder JM**, Senderek J. Towards a functional pathology of hereditary neuropathies. *Acta Neuropathol.* 133(4): 493-515, 2017

Ceccon G, Lohmann P, Stoffels G, Judov N, Filss CP, Rapp M, Bauer E, Hamisch C, Ruge MI, Kocher M, Kuchelmeister K, **Sellhaus B**, Sabel M, Fink GR, Shah NJ, Langen KJ, Galldiks N. Dynamic O-(2-18F-fluoroethyl)-L-tyrosine positron emission tomography differentiates brain metastasis recurrence from radiation injury after radiotherapy. *Neuro Oncol.* 19(2): 281-288, 2017

Musall S, **Haiss F**, Weber B, von der Behrens W. Deviant Processing in the Primary Somatosensory Cortex. *Cereb Cortex.* 27(1): 863-876, 2017

Reimann J, Kohlschmidt N, Tolksdorf K, **Weis J**, Kuchelmeister K, Roos A. Muscle Pathology as a Diagnostic Clue to Allgrove Syndrome. *J Neuropathol Exp Neurol.* 76(5): 337-341, 2017

Hube L, Dohrn MF, Karsai G, Hirshman S, Van Damme P, Schulz JB, **Weis J**, Hornemann T, Claeys KG. Metabolic Syndrome, Neurotoxic 1-Deoxysphingolipids and Nervous Tissue Inflammation in Chronic Idiopathic Axonal Polyneuropathy (CIAP). *PLoS One.* 12(1): e0170583, 2017

Gama-Carvalho M, Garcia-Vaquero ML, Pinto FR, Besse F, **Weis J**, Voigt A, Schulz JB, De Las Rivas J. Linking Amyotrophic Lateral Sclerosis and Spinal Muscular Atrophy through RNA-transcriptome homeostasis: a genomics perspective. *J Neurochem.* 141(1): 12-30, 2017

Kriebel A, **Hodde D**, Kuenzel T, Engels J, **Brook G**, Mey J. Cell-free artificial implants of electrospun fibres in a three-dimensional gelatin matrix support sciatic nerve regeneration in vivo. *J Tissue Eng Regen Med.* 11(12): 3289-3304, 2017

Brauers E, **Roos A**, Kollipara L, Zahedi RP, Beckmann A, Mohanadas N, Bauer H, Häusler M, Thoma S, Kress W, Senderek J, **Weis J**. The Caveolin-3 G56S sequence variant of unknown significance: Muscle biopsy findings and functional cell biological analysis. *Proteomics Clin Appl.* 11(1-2), 2017

van Neerven SG, Haastert-Talini K, Boecker A, Schriever T, Dabhi C, Claeys K, Deumens R, **Brook GA**, **Weis J**, Pallua N, Bozkurt A. Two-component collagen nerve guides support axonal regeneration in the rat peripheral nerve injury model. *J Tissue Eng Regen Med.* 11(12): 3349-3361, 2017

Veldeman M, Coburn M, Rossaint R, Clusmann H, **Nolte K**, Kremer B, Höllig A. Xenon Reduces Neuronal Hippocampal Damage and Alters the Pattern of Microglial Activation after Experimental Subarachnoid Hemorrhage: A Randomized Controlled Animal Trial. *Front Neurol.* 8: 511, 2017

Brücken A, Bleilevens C, Föhr P, **Nolte K**, Rossaint R, Marx G, Fries M, Derwall M. Influence of argon on temperature modulation and neurological outcome in hypothermia treated rats following cardiac arrest. *Resuscitation.* 117: 32-39, 2017

Dohrn MF, Glöckle N, Mulahasanovic L, Heller C, Mohr J, Bauer C, Riesch E, Becker A, Battke F, Hörtnagel K, Hornemann T, Suriyanarayanan S, Blankenburg M, Schulz JB, Claeys KG, Gess B, **Katona I**, Ferbert A, Vittore D, Grimm A, Wolking S, Schöls L, Lerche H, Korenke GC, Fischer D, Schrank B, Kotzaeridou U, Kurlermann G, Dräger B, Schirmacher A, Young P, Schlotter-Weigel B, Biskup S. Frequent genes in rare diseases: panel-based next generation sequencing to disclose causal mutations in hereditary neuropathies. *J Neurochem.* 143(5): 507-522, 2017

Doyen PJ, Vergouts M, Pochet A, Desmet N, van Neerven S, **Brook G**, Hermans E. Inflammation-associated regulation of RGS in astrocytes and putative implication in neuropathic pain. *J Neuroinflammation.* 14(1): 209, 2017

Publications 2016

Stenzel W, **Weis J**. Update on diagnostic muscle biopsy of neuromuscular diseases. *Nervenheilkunde*. 36: 17-22, 2016

Deschauer M, Mueller-Reible CR, Roesler KM, Schoser B, Wanschitz J, **Weis J**, Zierz S. Diagnosis of Myopathies. *Aktuelle Neurologie*. 43(10): 599-607, 2016

Kabelitz L, Nonn A, **Nolte KW**, Nikoubashman O, Othman A, Heringer S, Kramer M, Wiesmann M, Brockmann MA. Long Term Outcome after Application of the Angio-Seal Vascular Closure Device in Minipigs. *PLoS One*. 11(9): e0163878, 2016

Buchkremer S, González Coraspe JA, **Weis J**, Roos A. Sil1-Mutant Mice Elucidate Chaperone Function in Neurological Disorders. *J Neuromuscul Dis*. 3(2): 169-181, 2016

Bouhy D, Geuens T, De Winter V, Almeida-Souza L, **Katona I**, **Weis J**, Hocheleid T, Goossens S, Haigh JJ, Janssens S, Timmerman V. Characterization of New Transgenic Mouse Models for Two Charcot-Marie-Tooth-Causing HspB1 Mutations using the Rosa26 Locus. *J Neuromuscul Dis*. 3(2): 183-200, 2016

Altinova H, Möllers S, Deumens R, **Gerardo-Nava J**, Führmann T, van Neerven SG, Bozkurt A, Mueller CA, Hoff HJ, Heschel I, **Weis J**, **Brook GA**. Functional Recovery Not Correlated with Axon Regeneration through Olfactory Ensheathing Cell-Seeded Scaffolds in a Model of Acute Spinal Cord Injury. *Tissue Eng Regen Med*. 13(5): 585-600, 2016

Cordts I, **Funk F**, Schulz JB, **Weis J**, **Claeys KG**. Tubular aggregates in autoimmune Lambert-Eaton myasthenic syndrome. *Neuromuscul Disord*. 26(12): 880-884, 2016

Unger A, Dekomien G, Güttches A, Dreps T, Kley R, Tegenthoff M, Ferbert A, **Weis J**, Heyer C, Linke WA, Martinez-Carrera L, Storbeck M, Wirth B, Hoffjan S, Vorgerd M. Expanding the phenotype of BICD2 mutations toward skeletal muscle involvement. *Neurology*. 87(21): 2235-2243, 2016

Breuer T, Hatam N, Grabiger B, Marx G, Behnke BJ, **Weis J**, Kopp R, Gayan-Ramirez G, Zoremba N, Bruells CS. Kinetics of ventilation-induced changes in diaphragmatic metabolism by bilateral phrenic pacing in a piglet model. *Sci Rep*. 6: 35725, 2016

Bruells CS, Breuer T, Maes K, Bergs I, Bleilevens C, Marx G, **Weis J**, Gayan-Ramirez G, Rossaint R. Influence of weaning methods on the diaphragm after mechanical ventilation in a rat model. *BMC Pulm Med*. 16(1): 127, 2016

Joshi AR, Holtmann L, Bobylev I, Schneider C, Ritter C, **Weis J**, Lehmann HC. Loss of Schwann cell plasticity in chronic inflammatory demyelinating polyneuropathy (CIDP). *J Neuroinflammation*. 13(1): 255, 2016

Ferbort A, Zibat A, Rautenstrauss B, Kress W, Hügens-Penzel M, **Weis J**, Shah Y, Roth C. Laing distal myopathy with a novel mutation in exon 34 of the MYH7 gene. *Neuromuscul Disord*. 26(9): 598-603, 2016

Madeo M, Stewart M, Sun Y, Sahir N, Wiethoff S, Chandrasekar I, Yarrow A, Rosenfeld JA, Yang Y, Cordeiro D, McCormick EM, Muresku CC, Jepperson TN, McBeth LJ, Seidahmed MZ, El Khashab HY, Hamad M, **Azzedine H**, Clark K, Corrochano S, Wells S, Elting MW, Weiss MM, Burn S, Myers A, Landsverk M, Crotwell PL, Waisfisz Q, Wolf NI, Nolan PM, Padilla-Lopez S, Houlden H, Lifton R, Mane

S, Singh BB, Falk MJ, Mercimek-Mahmutoglu S, Bilguvar K, Salih MA, Acevedo-Arozena A, Kruer MC. Loss-of-Function Mutations in FRRS1L Lead to an Epileptic-Dyskinetic Encephalopathy. *Am J Hum Genet.* 98(6): 1249-55, 2016

Roos A, Kollipara L, **Buchkremer S**, **Labisch T**, **Brauers E**, **Gatz C**, **Lentz C**, **Gerardo-Nava J**, **Weis J**, Zahedi RP. Cellular Signature of SIL1 Depletion: Disease Pathogenesis due to Alterations in Protein Composition Beyond the ER Machinery. *Mol Neurobiol.* 53(8): 5527-41, 2016

Wolf HH, Kornhuber ME, **Weis J**, Posa A. Dysautonomic polyneuropathy as a variant of chronic inflammatory "demyelinating" polyneuropathy? *Clin Auton Res.* 26(4): 303-5; 2016

De Paepe B, Martin JJ, Herbelet S, Jimenez-Mallebrera C, Iglesias E, Jou C, **Weis J**, De Bleecker JL. Activation of osmolyte pathways in inflammatory myopathy and Duchenne muscular dystrophy points to osmoregulation as a contributing pathogenic mechanism. *Lab Invest.* 96(8): 872-84, 2016

Schrempf W, **Katona I**, Dogan I, Felbert VV, Wienecke M, Heller J, Maier A, Hermann A, Linse K, Brandt MD, Reichmann H, Schulz JB, Schiefer J, Oertel WH, Storch A, **Weis J**, Reetz K. Reduced intraepidermal nerve fiber density in patients with REM sleep behavior disorder. *Parkinsonism Relat Disord.* 29: 10-6, 2016

Werheid F, **Azzedine H**, Zwerenz E, Bozkurt A, Moeller MJ, Lin L, Mull M, Häusler M, Schulz JB, **Weis J**, **Claeys KG**. Underestimated associated features in CMT neuropathies: clinical indicators for the causative gene? *Brain Behav.* 6(4): e00451, 2016

Kollipara L, **Buchkremer S**, **Weis J**, **Brauers E**, Hoss M, Rütten S, Caviedes P, Zahedi RP, **Roos A**. Proteome Profiling and Ultrastructural Characterization of the Human RCMH Cell Line: Myoblastic Properties and Suitability for Myopathological Studies. *J Proteome Res.* 15(3): 945-55, 2016

Braczynski AK, Harter PN, Zeiner PS, Drott U, Tews DS, Preusse C, Penski C, Dunst M, **Weis J**, Stenzel W, Mittelbronn M. C5b-9 deposits on endomysial capillaries in non-dermatomyositis cases. *Neuromuscul Disord.* 26(4-5): 283-91, 2016

Claeys KG, Abicht A, Häusler M, Kleinle S, Wiesmann M, Schulz JB, Horvath R, **Weis J**. Novel genetic and neuropathological insights in NARP. *Muscle Nerve.* 54(2): 328-333, 2016

Rudnik-Schöneborn S, Deden F, Eggermann K, Eggermann T, Wieczorek D, **Sellhaus B**, **Yamoah A**, **Goswami A**, **Claeys KG**, **Weis J**, Zerres K. Autosomal dominant spinal muscular atrophy with lower extremity predominance: A recognizable phenotype of BICD2 mutations. *Muscle Nerve.* 54(3): 496-500, 2016

Altmann J, Büchner B, Nadaj-Pakleza A, Schäfer J, Jackson S, Lehmann D, Deschauer M, Kopajtich R, Lautenschläger R, Kuhn KA, Karle K, Schöls L, Schulz JB, **Weis J**, Prokisch H, Kornblum C, **Claeys KG**, Klopstock T. Expanded phenotypic spectrum of the m.8344A>G "MERRF" mutation: data from the German mitoNET registry. *J Neurol.* 263(5): 961-72, 2016

Ruegsegger C, Maharjan N, **Goswami A**, de L'Etang AF, **Weis J**, Troost D, Heller M, Gut H, Saxena S. Aberrant association of misfolded SOD1 with Na(+)/K(+)ATPase- α 3 impairs its activity and contributes to motor neuron vulnerability in ALS. *Acta Neuropathol.* 131(3): 427-51, 2016

Boecker AH, van Neerven SG, Scheffel J, Tank J, **Altinova H**, Seidensticker K, Deumens R, Tolba R, **Weis J**, **Brook GA**, Pallua N, Bozkurt A. Pre-differentiation of mesenchymal stromal cells in combination with a microstructured nerve guide supports peripheral nerve regeneration in the rat sciatic nerve model. *Eur J Neurosci.* 43(3): 404-16, 2016

Hodde D, Gerardo-Nava J, Wöhlk V, Weinandy S, Jockenhövel S, Kriebel A, **Altinova H**, Steinbusch HW, Möller M, **Weis J**, Mey J, **Brook GA**. Characterisation of cell-substrate interactions between Schwann cells and three-dimensional fibrin hydrogels containing orientated nanofibre topographical cues. *Eur J Neurosci*. 43(3): 376-87, 2016

Tauber SC, Staszewski O, Prinz M, **Weis J**, **Nolte K**, Bunkowski S, Brück W, Nau R. HIV encephalopathy: glial activation and hippocampal neuronal apoptosis, but limited neural repair. *HIV Med*. 17(2): 143-51, 2016

Röhrich M, Koelsche C, Schrimpf D, Capper D, Sahm F, Kratz A, Reuss J, Hovestadt V, Jones DT, Bewerunge-Hudler M, Becker A, **Weis J**, Mawrin C, Mittelbronn M, Perry A, Mautner VF, Mecht-ersheimer G, Hartmann C, Okuducu AF, Arp M, Seiz-Rosenhagen M, Hänggi D, Heim S, Paulus W, Schittenhelm J, Ahmadi R, Herold-Mende C, Unterberg A, Pfister SM, von Deimling A, Reuss DE. Methylation-based classification of benign and malignant peripheral nerve sheath tumors. *Acta Neuro-pathol*. 131(6): 877-87, 2016

Bozkurt A, Boecker A, Tank J, **Altinova H**, Deumens R, Dabhi C, Tolba R, **Weis J**, **Brook GA**, Pallua N, van Neerven SG. Efficient bridging of 20 mm rat sciatic nerve lesions with a longitudinally micro-structured collagen scaffold. *Biomaterials*. 75: 112-22, 2016

Publications 2015

Güttsches AK, Dekomien G, **Claeys KG**, von der Hagen M, Huebner A, Kley RA, Kirschner J, Vorgerd M. Two novel nebulin variants in an adult patient with congenital nemaline myopathy. *Neuromuscul Disord*. 25(5): 392-6, 2015

Musumeci O, Thieme A, **Claeys KG**, Wenninger S, Kley RA, Kuhn M, Lukacs Z, Deschauer M, Gaeta M, Toscano A, Gläser D, Schoser B. Homozygosity for the common GAA gene splice site mutation c.-32-13T>G in Pompe disease is associated with the classical adult phenotypical spectrum. *Neuromuscul Disord*. 25(9): 719-24, 2015

Bernard-Marissal N, Médard JJ, **Azzedine H**, Chrast R. Reply: Is SIGMAR1 a confirmed FTD/MND gene? *Brain*. 138(Pt 11): e394, 2015

Khaminets A, Heinrich T, Mari M, Grumati P, Huebner AK, Akutsu M, Liebmann L, Stolz A, Nietzsche S, Koch N, Mauthe M, **Katona I**, Qualmann B, **Weis J**, Reggiori F, Kurth I, Hübner CA, Dikic I. Regulation of endoplasmic reticulum turnover by selective autophagy. *Nature*. 522 (7556): 354-8, 2015

Mayrhofer JM, **Haiss F**, Haenni D, Weber S, Zuend M, Barrett MJ, Ferrari KD, Maechler P, Saab AS, Stobart JL, Wyss MT, Johannssen H, Osswald H, Palmer LM, Revol V, Schuh CD, Urban C, Hall A, Larkum ME, Rutz-Innerhofer E, Zeilhofer HU, Ziegler U, Weber B. Design and performance of an ultra-flexible two-photon microscope for in vivo research. *Biomed Opt Express*. 6(11): 4228-37, 2015

Mayrhofer JM, **Haiss F**, Helmchen F, Weber B. Sparse, reliable, and long-term stable representation of periodic whisker deflections in the mouse barrel cortex. *Neuroimage*. 115: 52-63, 2015

Höllig A, **Weinandy A**, **Nolte K**, Clusmann H, Rossaint R, Coburn M. Experimental subarachnoid hemorrhage in rats: comparison of two endovascular perforation techniques with respect to success rate, confounding pathologies and early hippocampal tissue lesion pattern. *PLoS One*. 10(4): e0123398, 2015

König LS, Wiesmann M, Pjontek R, **Sellhaus B**, Schulz JB, Tauber SC. Amyloid beta-related angiitis as rare cause of a generalized convulsive seizure. *Nervenarzt*. 86(10): 1270-2, 2015

Metz I, Traffehn S, Straßburger-Krogias K, Keyvani K, Bergmann M, **Nolte K**, Weber MS, Bartsch T, Gold R, Brück W. Glial cells express nuclear nrf2 after fumarate treatment for multiple sclerosis and psoriasis. *Neurol Neuroimmunol Neuroinflamm*. 2(3): e99, 2015

Leipold E, Hanson-Kahn A, Frick M, Gong P, Bernstein JA, Voigt M, **Katona I**, Oliver Goral R, Altmüller J, Nürnberg P, **Weis J**, Hübner CA, Heinemann SH, Kurth I. Cold-aggravated pain in humans caused by a hyperactive NaV1.9 channel mutant. *Nature Commun*. 6: 10049, 2015

Brücken A, Derwall M, Bleilevens C, Stoppe C, Götzenich A, Gaisa NT, **Weis J**, **Nolte KW**, Rossaint R, Ichinose F, Fries M. Brief inhalation of nitric oxide increases resuscitation success and improves 7-day-survival after cardiac arrest in rats: a randomized controlled animal study. *Crit Care*. 19: 408, 2015

Thron A, Krings T, Otto J, Mull M, **Schroeder JM**. The Transdural Course of Radicular Spinal Cord Veins-A Microangiographical and Microscopical Study. *Clin Neuroradiol*. 25(4): 361-9, 2015

Derwall M, Ebeling A, **Nolte KW**, **Weis J**, Rossaint R, Ichinose F, Nix C, Fries M, Brücken A. Inhaled nitric oxide improves transpulmonary blood flow and clinical outcomes after prolonged cardiac arrest: a large animal study. *Crit Care*. 19: 328, 2015

Maier A, Mannartz V, Wasmuth H, Trautwein C, Neumann UP, **Weis J**, Grosse J, Fuest M, Hilz MJ, Schulz JB, Haubrich C. GAD Antibodies as Key Link Between Chronic Intestinal Pseudoobstruction, Autonomic Neuropathy, and Limb Stiffness in a Nondiabetic Patient: A CARE-Compliant Case Report and Review of the Literature. *Medicine (Baltimore)*. 94(31): e1265, 2015

Abagnale G, Steger M, Nguyen VH, Hersch N, Sechi A, Jousen S, Denecke B, Merkel R, Hoffmann B, **Dreser A**, Schnakenberg U, Gillner A, Wagner W. Surface topography enhances differentiation of mesenchymal stem cells towards osteogenic and adipogenic lineages. *Biomaterials*. 61: 316-26, 2015

Johann S, Heitzer M, Kanagaratnam M, **Goswami A**, Rizo T, **Weis J**, Troost D, Beyer C. NLRP3 inflammasome is expressed by astrocytes in the SOD1 mouse model of ALS and in human sporadic ALS patients. *Glia*. 63(12): 2260-73, 2015

Kronenbuerger M, **Nolte KW**, Coenen VA, Burgunder JM, Krauss JK, **Weis J**. Brain alterations with deep brain stimulation: New insight from a neuropathological case series. *Mov Disord*. 30(8): 1125-30, 2015

Chen YC, Auer-Grumbach M, Matsukawa S, Zitzelsberger M, Themistocleous AC, Strom TM, Samara C, Moore AW, Cho LT, Young GT, Weiss C, Schabhüttl M, Stucka R, Schmid AB, Parman Y, Graul-Neumann L, Heinritz W, Passarge E, Watson RM, Hertz JM, Moog U, Baumgartner M, Valente EM, Pereira D, Restrepo CM, **Katona I**, Dusl M, Stendel C, Wieland T, Stafford F, Reimann F, von Au K, Finke C, Willems PJ, Nahorski MS, Shaikh SS, Carvalho OP, Nicholas AK, Karbani G, McAleer MA, Cilio MR, McHugh JC, Murphy SM, Irvine AD, Jensen UB, Windhager R, **Weis J**, Bergmann C, Rautenstrauss B, Baets J, De Jonghe P, Reilly MM, Kropatsch R, Kurth I, Chrast R, Michiue T, Bennett DL, Woods CG, Senderek J. Transcriptional regulator PRDM12 is essential for human pain perception. *Nature Genet*. 47(7): 803-8, 2015

Bernard-Marissal N, Médard JJ, **Azzedine H***, Chrast R*. Dysfunction in endoplasmic reticulum-mitochondria crosstalk underlies SIGMAR1 loss of function mediated motor neuron degeneration. *Brain*. 138: 875-90, 2015 *Equal contribution

Baets J, Duan X, Wu Y, Smith G, Seeley WW, Mademan I, McGrath NM, Beadell NC, Khoury J, Botuyan MV, Mer G, Worrell GA, Hojo K, DeLeon J, Laura M, Liu YT, Senderek J, **Weis J**, Van den Bergh P, Merrill SL, Reilly MM, Houlden H, Grossman M, Scherer SS, De Jonghe P, Dyck PJ, Klein CJ. Defects of mutant DNMT1 are linked to a spectrum of neurological disorders. *Brain*. 138: 845-61, 2015

Brücken A, Kurnaz P, Bleilevens C, Derwall M, **Weis J**, **Nolte K**, Rossaint R, Fries M. Delayed Argon Administration Provides Robust Protection Against Cardiac Arrest-Induced Neurological Damage. *Neurocrit Care*. 22(1): 112-20, 2015

Roos A, **Weis J**, Korinthenberg R, Fehrenbach H, Häusler M, Züchner S, Mache C, Hubmann H, Auer-Grumbach M, Senderek J. Inverted formin 2-related Charcot-Marie-Tooth disease: extension of the mutational spectrum and pathological findings in Schwann cells and axons. *J Peripher Nerv Syst*. 20(1): 52-9, 2015

Ruschel J, Hellal F, Flynn KC, Dupraz S, Elliott DA, Tedeschi A, Bates M, Sliwinski C, **Brook G**, Dobrint K, Peitz M, Brüstle O, Norenberg MD, Blesch A, Weidner N, Bunge MB, Bixby JL, Bradke F. Axonal regeneration. Systemic administration of epothilone B promotes axon regeneration after spinal cord injury. *Science*. 348(6232): 347-52, 2015

Stenzel W, Preuße C, Allenbach Y, Pehl D, Junckerstorff R, Heppner FL, **Nolte K**, Aronica E, Kana V, Rushing E, Schneider U, **Claeys KG**, Benveniste O*, **Weis J***, Goebel HH*. Nuclear actin aggregation is a hallmark of anti-synthetase syndrome-induced dysimmune myopathy. *Neurology*. 84(13): 1346-54, 2015 *Equal contribution

Dohrn MF, Othman A, Hirshman SK, Bode H, Alecu I, Fähndrich E, Karges W, **Weis J**, Schulz JB, Hornemann T, **Claeys KG**. Elevation of plasma 1-deoxy-sphingolipids in type 2 diabetes mellitus: a susceptibility to neuropathy? *Eur J Neurol*. 22(5): 806-e55, 2015

Filézac de L'Etang A, Maharjan N, Cordeiro Braña M, Ruegsegger C, Rehmann R, **Goswami A**, Roos A, Troost D, Schneider BL, **Weis J**, Saxena S. Marinesco-Sjögren syndrome protein SIL1 regulates motor neuron subtype-selective ER stress in ALS. *Nature Neurosci*. 18(2): 227-38, 2015

Goswami A, **Jesse C**, **Chandrasekar A**, **Bushuven E**, **Vollrath J**, **Dreser A**, **Katona I**, Beyer C, Johann S, Feller A, Grond M, Wagner S, **Nikolin S**, Troost D, **Weis J**. Accumulation of STIM1 is associated with the degenerative muscle fibre phenotype in ALS and other neurogenic atrophies. *Neuropathol Appl Neurobiol*. 41(3): 304-18, 2015

Publications 2014

Peripheral nerve disorders: pathology and genetics / edited by Jean-Michel Vallat, **Joachim Weis**. The International Society of Neuropathology; *Wiley Blackwell* ISBN: 978-1-118-61843-1, 2014

Bozkurt A, Apel C, **Sellhaus B**, van Neerven S, Wessing B, Hilgers RD, Pallua N. Differences in degradation behavior of two non-cross-linked collagen barrier membranes: an in vitro and in vivo study. *Clin Oral Implants Res*. 25(12): 1403-11, 2014

Gerardo-Nava J, **Hodde D**, **Katona I**, Bozkurt A, Grehl T, Steinbusch HWM, **Weis J**, **Brook GA**. The growth and regeneration of axons: an in vitro model for studying interactions in 3D *Folia Neuropathologica* 52: 307-8, 2014

Schelleckes M, Lenders M, Guske K, Schmitz B, Tanislav C, Ständer S, Metze D, **Katona I**, **Weis J**, Brand SM, Duning T, Brand E. Cryptogenic stroke and small fiber neuropathy of unknown etiology in

patients with alpha-galactosidase A -10T genotype. *Orphanet J Rare Dis.* 9(1): 178, 2014

Böhm J, Biancalana V, Malfatti E, Dondaine N, Koch C, Vasli N, Kress W, Strittmatter M, Taratuto AL, Gonorazky H, Laforêt P, Maissonobe T, Olivé M, Gonzalez-Mera L, Fardeau M, Carrière N, Clavelou P, Eymard B, Bitoun M, Rendu J, Fauré J, **Weis J**, Mandel JL, Romero NB, Laporte J. Adult-onset autosomal dominant centronuclear myopathy due to BIN1 mutations. *Brain.* 137(Pt 12): 3160-70, 2014

Musall S, von der Behrens W, Mayrhofer JM, Weber B, Helmchen F, **Haiss F**. Tactile frequency discrimination is enhanced by circumventing neocortical adaptation. *Nature Neurosci.* 17(11): 1567-73, 2014

Semmler AL, Sacconi S, Bach J, **Liebe C**, Bürmann J, Kley RA, Ferbert A, Anderheiden R, Van den Bergh P, Martin JJ, De Jonghe P, Neuen-Jacob E, Müller O, Deschauer M, Bergmann M, Schröder J, Vorgerd M, Schulz JB, **Weis J**, Kress W, **Claeys KG**. Unusual multisystemic involvement and a novel BAG3 mutation revealed by NGS screening in a large cohort of myofibrillar myopathies. *Orphanet J Rare Dis.* 9(1): 121, 2014

Altinova H, Möllers S, Führmann T, Deumens R, Bozkurt A, Heschel I, Damink LH, Schügner F, **Weis J**, **Brook GA**. Functional improvement following implantation of a microstructured, type-I collagen scaffold into experimental injuries of the adult rat spinal cord. *Brain Res.* 1585: 37-50, 2014

Müller TJ, Kraya T, Stoltenburg-Didingen G, Hanisch F, Kornhuber M, Stoevesandt D, Senderek J, **Weis J**, Baum P, Deschauer M, Zierz S. Phenotype of Matrin 3 related distal myopathy in 16 German patients. *Ann Neurol.* 76(5): 669-80, 2014

Vollrath JT, Sechi A, **Dreser A**, **Katona I**, Wiemuth D, Vervoorts J, Dohmen M, **Chandrasekar A**, **Prause J**, **Brauers E**, **Jesse CM**, **Weis J***, **Goswami A***. Loss of function of the ALS protein SigR1 leads to ER pathology associated with defective autophagy and lipid raft disturbances. *Cell Death Dis.* 5: e1290, 2014 ; *equal contribution

Bozkurt A, van Neerven SG, **Claeys KG**, O'Dey DM, Sudhoff A, **Brook GA**, **Sellhaus B**, Schulz JB, **Weis J**, Pallua N. The proximal medial sural nerve biopsy model: a standardised and reproducible baseline clinical model for the translational evaluation of bioengineered nerve guides. *Biomed Res Int.* 2014:121452, 2014

van Neerven SG, Krings L, Haastert-Talini K, Vogt M, Tolba RH, **Brook G**, Pallua N, Bozkurt A. Human Schwann cells seeded on a novel collagen-based microstructured nerve guide survive, proliferate, and modify neurite outgrowth. *Biomed Res Int.* 2014:493823, 2014

Elbracht M, Senderek J, Schara U, **Nolte K**, Klopstock T, **Roos A**, Reimann J, Zerres K, **Weis J**, Rudnik-Schöneborn S. Clinical and morphological variability of the E396K mutation in the neurofilament light chain gene in patients with Charcot-Marie-Tooth disease type 2E. *Clin Neuropathol.* 33(5): 335-43, 2014

Roos A, **Buchkremer S**, Kollipara L, **Labisch T**, **Gatz C**, Zitzelsberger M, **Brauers E**, **Nolte K**, **Schröder JM**, Kirschner J, **Jesse CM**, Goebel HH, **Goswami A**, Zimmermann R, Zahedi RP, Senderek J, **Weis J**. Myopathy in Marinesco-Sjögren syndrome links endoplasmic reticulum chaperone dysfunction to nuclear envelope pathology. *Acta Neuropathol.* 127(5): 761-77, 2014

Del Bigio MR, Hainfellner JA, McLean CA, Powell SZ, Sikorska B, Takahashi H, **Weis J**, Xuereb JH. Neuropathology training worldwide - evolution and comparisons. *Brain Pathol.* 24(3): 285-98, 2014

Weinandy A, Piroth MD, **Goswami A**, **Nolte K**, **Sellhaus B**, Gerardo-Nava J, Eble M, Weinandy S, Cornelissen C, Clusmann H, Lüscher B, **Weis J**. Cetuximab induces eme1-mediated DNA repair: a novel mechanism for cetuximab resistance. *Neoplasia*. 16(3): 207-20, 2014

Brücken A, Kurnaz P, Bleilevens C, Derwall M, **Weis J**, **Nolte K**, Rossaint R, Fries M. Dose dependent neuroprotection of the noble gas argon after cardiac arrest in rats is not mediated by K(ATP)-Channel opening. *Resuscitation*. 85(6): 826-32, 2014

Djémié T, Weckhuysen S, Holmgren P, Hardies K, Van Dyck T, Hendrickx R, Schoonjans AS, Van Paesschen W, Jansen AC, De Meirleir L, Selim LA, Girgis MY, Buyse G, Lagae L, Smets K, Smouts I, **Claeys KG**, Van den Bergh V, Grisar T, Blatt I, Shorer Z, Roelens F, Afawi Z, Helbig I, Ceulemans B, De Jonghe P, Suls A. PRRT2 mutations: exploring the phenotypical boundaries. *J Neurol Neurosurg Psychiatry*. 85(4): 462-5, 2014

Flöck A, Kornblum C, Hammerstingl C, **Claeys KG**, Gembruch U, Merz WM. Progressive cardiac dysfunction in Bethlehem myopathy during pregnancy. *Obstet Gynecol*. 123(2 Pt 2 Suppl 2): 436-8, 2014

Haensch CA, Tosch M, **Katona I**, **Weis J**, Isenmann S. Small-fiber neuropathy with cardiac denervation in postural tachycardia syndrome. *Muscle Nerve*. 50(6): 956-61, 2014

Gerardo-Nava J, **Hodde D**, **Katona I**, Bozkurt A, Grehl T, Steinbusch HW, **Weis J**, **Brook GA**. Spinal cord organotypic slice cultures for the study of regenerating motor axon interactions with 3D scaffolds. *Biomaterials*. 35(14): 4288-96, 2014

Kriebel A, Rumman M, Scheld M, **Hodde D**, **Brook G**, Mey J. Three-dimensional configuration of orientated fibers as guidance structures for cell migration and axonal growth. *J Biomed Mater Res B Appl Biomater*. 102(2): 356-65, 2014

Joshi PR, Hauburger A, Kley R, **Claeys KG**, Schneider I, Kress W, Stoltenburg G, **Weis J**, Vorgerd M, Deschauer M, Hanisch F. Mitochondrial abnormalities in myofibrillar myopathies. *Clin Neuropathol*. 33(2): 134-42, 2014

Schreckenbach T, **Schröder JM**, Voit T, Abicht A, Neuen-Jacob E, **Roos A**, Bulst S, Kuhl C, Schulz JB, **Weis J**, **Claeys KG**. Novel TPM3 mutation in a family with cap myopathy and review of the literature. *Neuromuscul Disord*. 24(2): 117-24, 2014

Katona I, **Weis J**, Hanisch F. Glycogenosome accumulation of the arrector pili muscle in Pompe disease. *Orphanet J Rare Dis*. 9(1): 17, 2014

Bruells CS, Bergs I, Rossaint R, Du J, Bleilevens C, Goetzenich A, **Weis J**, Wiggs MP, Powers SK, Hein M. Recovery of Diaphragm Function following Mechanical Ventilation in a Rodent Model. *PLoS One*. 9(1): e87460, 2014

Linz U, Ulus B, Neuloh G, Clusmann H, Oertel M, **Nolte K**, **Weis J**, Heussen N, Gilsbach JM. Can in-vitro chemoresponse assays help find new treatment regimens for malignant gliomas? *Anticancer Drugs*. 25(4): 375-84, 2014

Joshi PR, Gläser D, Dreßel C, Kress W, **Weis J**, Deschauer M. Anoctamin 5 muscular dystrophy associated with a silent p.Leu115Leu mutation resulting in exon skipping. *Neuromuscul Disord*. 24(1): 43-7, 2014

Poretti A, Häusler M, von Moers A, Baumgartner B, Zerres K, Klein A, Aiello C, Moro F, Zanni G, Santorelli FM, Huisman TA, **Weis J**, Valente EM, Bertini E, Boltshauser E. Ataxia, Intellectual Disability, and Ocular Apraxia with Cerebellar Cysts: A New Disease? *Cerebellum*. 13(1): 79-88, 2014

Wild F, Tuettenberg J, Grau A, **Weis J**, Krauss JK. Ligamentum flavum hematomas of the cervical and thoracic spine. *Clin Neurol Neurosurg*. 116: 24-7, 2014

Bruells CS, Maes K, Rossaint R, Thomas D, Cielen N, Bergs I, Bleilevens C, **Weis J**, Gayan-Ramirez G. Sedation Using Propofol Induces Similar Diaphragm Dysfunction and Atrophy during Spontaneous Breathing and Mechanical Ventilation in Rats. *Anesthesiology*. 120(3):665-72, 2014

Tauber SC, Harms K, Falkenburger B, **Weis J**, **Sellhaus B**, Nau R, Schulz JB, Reich A. Modulation of Hippocampal Neuroplasticity by Fas/CD95 Regulatory Protein 2 (Faim2) in the Course of Bacterial Meningitis. *J Neuropathol Exp Neurol*. 73(1): 2-13, 2014

Brook GA, **Hodde D**, Kretschmer T. Grundlegendes zur Degeneration und Regeneration von Nerven. In T. Kretschmer, G. Antoniadis, H. Assmus (Hrsg.): Nerven Chirurgie. ISBN: 978-3-642-36894-3. *Springer* 1-10, 2014

Heinen C, Kretschmer T, **Weis J**. Nerventumoren. In T. Kretschmer, G. Antoniadis, H. Assmus (Hrsg.): Nerven Chirurgie. ISBN: 978-3-642-36894-3. *Springer* 227-59, 2014

Publications 2013

Schreckenbach T, Henn W, Kress W, **Roos A**, Maschke M, Feiden W, Dillmann U, Schulz JB, **Weis J**, **Claeys KG**. Novel FHL1 mutation in a family with reducing body myopathy. *Muscle Nerve*. 47(1): 127-34, 2013

Prause J, **Goswami A**, **Katona I**, **Roos A**, Schnitzler M, **Bushuven E**, **Dreier A**, **Buchkremer S**, Johann S, Beyer C, Deschauer M, Troost D, **Weis J**. Altered localization, abnormal modification and loss of function of Sigma receptor-1 in amyotrophic lateral sclerosis. *Hum Mol Genet*. 22(8): 1581-600, 2013

Nolte KW, Trepels-Kotteck S, Honnef D, **Weis J**, Bien CG, van Baalen A, Ritter K, Czermin B, Rudnik-Schöneborn S, Wagner N, Häusler M. Early muscle and brain ultrastructural changes in polymerase gamma 1-related encephalomyopathy. *Neuropathology*. 33(1): 59-67, 2013

Brücken A, Cizen A, Fera C, Meinhardt A, **Weis J**, **Nolte K**, Rossaint R, Pufe T, Marx G, Fries M. Argon reduces neurohistopathological damage and preserves functional recovery after cardiac arrest in rats. *Br J Anaesth*. 110 Suppl 1: i106-i112, 2013

Schulz A, Baader SL, Niwa-Kawakita M, Jung MJ, Bauer R, Garcia C, Zoch A, Schacke S, Hagel C, Mautner VF, Hanemann CO, Dun XP, Parkinson DB, **Weis J**, **Schröder JM**, Gutmann DH, Giovannini M, Morrison H. Merlin isoform 2 in neurofibromatosis type 2-associated polyneuropathy. *Nat Neurosci*. 16(4): 426-33, 2013

Funk F, Ceuterick-de Groote C, Martin JJ, **Meinhardt A**, Taratuto AL, De Bleecker J, Van Coster R, De Paepe B, Schara U, Vorgerd M, Häusler M, Koppi S, Maschke M, De Jonghe P, Van Maldergem L, Noel S, Zimmermann CW, Wirth S, Isenmann S, Stadler R, **Schröder JM**, Schulz JB, **Weis J**, **Claeys KG**. Morphological spectrum and clinical features of myopathies with tubular aggregates. *Histol Histopathol*. 28(8): 1041-54, 2013

Böhm J, Vasli N, Maurer M, Cowling B, Shelton GD, Kress W, Toussaint A, Prokic I, Schara U, Anderson TJ, **Weis J**, Tired L, Laporte J. Altered Splicing of the BIN1 Muscle-Specific Exon in Humans and Dogs with Highly Progressive Centronuclear Myopathy. *PLoS Genet.* 9(6): e1003430, 2013

Ermis U, **Weis J**, Schulz JB. PML in a patient treated with fumaric acid. *N Engl J Med.* 368(17): 1657-8, 2013

Elsas J, **Sellhaus B**, Herrmann M, Kinkeldey A, **Weis J**, Jahnen-Dechent W, Häusler M. Fetuin-A in the developing brain. *Dev Neurobiol.* 73(5): 354-69, 2013

Azzedine H, Zavadakova P, Planté-Bordeneuve V, Vaz Pato M, Pinto N, Bartesaghi L, Zenker J, Poirot O, Bernard-Marissal N, Arnaud Gouttenoire E, Cartoni R, Title A, Venturini G, Médard JJ, Makowski E, Schöls L, **Claeys KG**, Stendel C, **Roos A**, **Weis J**, Dubourg O, Leal Loureiro J, Stevanin G, Said G, Amato A, Baraban J, Leguern E, Senderek J, Rivolta C, Chrast R. PLEKHG5 deficiency leads to an intermediate form of autosomal recessive Charcot-Marie-Tooth disease. *Hum Mol Genet.* 22(20): 4224-32, 2013

Bruells CS, Maes K, Rossaint R, Thomas D, Cielen N, Bleilevens C, Bergs I, Loetscher U, **Dreier A**, Gayan-Ramirez G, Behnke BJ, **Weis J**. Prolonged Mechanical Ventilation Alters the Expression Pattern of Angio-neogenetic Factors in a Pre-Clinical Rat Model. *PLoS One.* 8(8): e70524, 2013

Claeys KG, **Gorodinskaya O**, Handt S, Reimann J, Kress W, Kornblum C, Kuhl C, Schulz JB, **Weis J**. Diagnostic challenge and therapeutic dilemma in necrotizing myopathy. *Neurology.* 81(10): 932-5, 2013

Leipold E, Liebmann L, Korenke GC, Heinrich T, Gießelmann S, Baets J, Ebbinghaus M, Goral RO, Stöberg T, Hennings JC, Bergmann M, Altmüller J, Thiele H, Wetzel A, Nürnberg P, Timmerman V, De Jonghe P, Blum R, Schaible HG, **Weis J**, Heinemann SH, Hübner CA, Kurth I. A de novo gain-of-function mutation in SCN11A causes loss of pain perception. *Nature Genet.* 45(11): 1399-404, 2013

Ermis U, **Weis J**, Schulz JB. Case reports of PML in patients treated for psoriasis. *N Engl J Med.* 369(11): 1081, 2013

Brunn A, Nagel I, Montesinos-Rongen M, Klapper W, Vater I, Paulus W, Hans V, Blümcke I, **Weis J**, Siebert R, Deckert M. Frequent triple-hit expression of MYC, BCL2, and BCL6 in primary lymphoma of the central nervous system and absence of a favorable MYC(low)BCL2 (low) subgroup may underlie the inferior prognosis as compared to systemic diffuse large B cell lymphomas. *Acta Neuropathol.* 126(4): 603-5, 2013

Gerardo-Nava J, **Mayorenko II**, Grehl T, Steinbusch HW, **Weis J**, **Brook GA**. Differential pattern of neuroprotection in lumbar, cervical and thoracic spinal cord segments in an organotypic rat model of glutamate-induced excitotoxicity. *J Chem Neuroanat.* 53: 11-7, 2013

Salih MA, Mundwiler E, Khan AO, Aldrees A, Elmalik SA, Hassan HH, Al-Owain M, Alkhalidi HM, **Katona I**, Kabiraj MM, Chrast R, Kentab AY, Alzaidan H, Rodenburg RJ, Bosley TM, **Weis J**, Koenig M, Stevanin G, Azzedine H. New Findings in a Global Approach to Dissect the Whole Phenotype of PLA2G6 Gene Mutations. *PLoS One.* 8(10): e76831, 2013

Rana OR, Schröder JW, Baukloh JK, Saygili E, Mischke K, Schiefer J, **Weis J**, Marx N, Rassaf T, Kelm M, Shin DI, Meyer C, Saygili E. Neurofilament light chain as an early and sensitive predictor of long-term neurological outcome in patients after cardiac arrest. *Int J Cardiol.* 168(2): 1322-7, 2013

Krieger M, **Roos A**, Stendel C, **Claeys KG**, Sonmez FM, Baudis M, Bauer P, Bornemann A, de Goede C, Dufke A, Finkel RS, Goebel HH, Häussler M, Kingston H, Kirschner J, Medne L, Muschke P, Rivier F, Rudnik-Schöneborn S, Spengler S, Inzana F, Stanzial F, Benedicenti F, Synofzik M, Lia Taratuto A, Pirra L, Tay SK, Topaloglu H, Uyanik G, Wand D, Williams D, Zerres K, **Weis J**, Senderek J. SIL1 mutations and clinical spectrum in patients with Marinesco-Sjogren syndrome. *Brain*. 136(Pt 12): 3634-44, 2013

Dohrn MF, Röcken C, De Bleecker JL, Martin JJ, Vorgerd M, Van den Bergh PY, Ferbert A, Hinderhofer K, **Schröder JM**, **Weis J**, Schulz JB, **Claeys KG**. Diagnostic hallmarks and pitfalls in late-onset progressive transthyretin-related amyloid-neuropathy. *J Neurol*. 260(12): 3093-108, 2013

Hanisch F, Weidemann W, Großmann M, Joshi PR, Holzhausen HJ, Stoltenburg G, **Weis J**, Zierz S, Horstkorte R. Sialylation and muscle performance: sialic Acid is a marker of muscle ageing. *PLoS One*. 8(12): e80520, 2013

Hans FJ, Geibprassert S, Krings T, **Weis J**, Deckert M, Ludolph A, Osieka R, Jost E. Solitary Plasmacytoma Presenting as an Intramedullary Mass of the Cervical Cord. *J Neurol Surg A Cent Eur Neurosurg*. 74 Suppl 1: e13-7, 2013

Gerdjikov TV, **Haiss F**, Rodriguez-Sierra OE, Schwarz C. Rhythmic whisking area (RW) in rat primary motor cortex: an internal monitor of movement-related signals? *J Neurosci*. 33(35): 14193-204, 2013

Deumens R, Van Gorp SF, Bozkurt A, Beckmann C, Führmann T, Montzka K, Tolba R, Kobayashi E, Heschel I, **Weis J**, **Brook GA**. Motor outcome and allodynia are largely unaffected by novel olfactory ensheathing cell grafts to repair low-thoracic lesion gaps in the adult rat spinal cord. *Behav Brain Res*. 237: 185-9, 2013

Siegel S, Streetz-van der Werf C, Schott JS, **Nolte K**, Karges W, Kreitschmann-Andermahr I. Diagnostic delay is associated with psychosocial impairment in acromegaly. *Pituitary*. 16: 507-14, 2013

Cantinieux D, Quertainmont R, Blacher S, Rossi L, Wanet T, Noël A, **Brook G**, Schoenen J, Franzen R. Conditioned Medium from Bone Marrow-Derived Mesenchymal Stem Cells Improves Recovery after Spinal Cord Injury in Rats: An Original Strategy to Avoid Cell Transplantation. *PLoS One*. 8(8): e69515, 2013

Piroth MD, Prasath J, Willuweit A, Stoffels G, **Sellhaus B**, van Osterhout A, Geisler S, Shah NJ, Eble MJ, Coenen HH, Langen KJ. Uptake of O-(2-[F-18]fluoroethyl)-L-tyrosine in reactive astrocytosis in the vicinity of cerebral gliomas. *Nucl Med Biol*. 40(6): 795-800, 2013

Gouttenoire EA, Lupo V, Calpena E, Bartesaghi L, Schüpfer F, Médard JJ, Maurer F, Beckmann JS, **Senderek J**, Palau F, Espinós C, Chrast R. Sh3tc2 deficiency affects neuregulin-1/ErbB signalling. *Glia*. 61(7): 1041-51, 2013

Billot S, Hervé D, Akman HO, Froissart R, Baussan C, **Claeys KG**, Piraud M, Sedel F, Mochel F, Laforêt P. Acute but transient neurological deterioration revealing adult polyglucosan body disease. *J Neurol Sci*. 324(1-2): 179-82, 2013

Publications 2012

Saygili E, Rana OR, Günzel C, Rackauskas G, Saygili E, Noor-Ebad F, Gemein C, Zink MD, Schwinger RH, Mischke K, **Weis J**, Marx N, Schauerte P. Rate and irregularity of electrical activation during atrial

fibrillation affect myocardial NGF expression via different signalling routes. *Cell Signal*. 24(1): 99-105, 2012

Bozkurt A, Lassner F, O'Dey D, **Deumens R**, Böcker A, Schwendt T, Janzen C, Suschek CV, Tolba R, Kobayashi E, **Sellhaus B**, Tholl S, Eummelen L, Schügner F, Olde Damink L, **Weis J**, **Brook GA**, Pallua N. The role of microstructured and interconnected pore channels in a collagen-based nerve guide on axonal regeneration in peripheral nerves. *Biomaterials*. 33(5): 1363-75, 2012

Dreier A, Barth S, **Goswami A**, **Weis J**. Cetuximab induces mitochondrial translocalization of EGFRvIII, but not EGFR: involvement of mitochondria in tumor drug resistance? *Tumour Biol*. 33(1): 85-94, 2012

Parthey K, Kornhuber M, Kunze C, Wand D, **Nolte KW**, **Nikolin S**, **Weis J**, **Schröder JM**. SOX10 mutation with peripheral amyelination and developmental disturbance of axons. *Muscle Nerve*. 45(2): 284-90, 2012

De Paepe B, Creus KK, **Weis J**, De Bleecker JL. Heat shock protein families 70 and 90 in Duchenne muscular dystrophy and inflammatory myopathy: Balancing muscle protection and destruction. *Neuromuscul Disord*. 22(1): 26-33, 2012

Groh J, **Weis J**, Zieger H, Stanley ER, Heuer H, Martini R. Colony-stimulating factor-1 mediates macrophage-related neural damage in a model for Charcot-Marie-Tooth disease type 1X. *Brain*. 135(Pt 1): 88-104, 2012

Weis J, Brandner S, Lammens M, Sommer C, Vallat JM. Processing of nerve biopsies: A practical guide for neuropathologists. *Clin Neuropathol*. 31(1): 7-23, 2012

Rana OR, Schröder JW, Kühnen JS, Saygili E, Gemein C, Zink MD, Schauerte P, Schiefer J, Schwinger RH, **Weis J**, Marx N, Kelm M, Meyer C, Saygili E. The Modified Glasgow Outcome Score for the prediction of outcome in patients after cardiac arrest: a prospective clinical proof of concept study. *Clin Res Cardiol*. 101(7): 533-43, 2012

Béhin A, Jardel C, **Claeys KG**, Fagart J, Louha M, Romero NB, Laforêt P, Eymard B, Lombès A. Adult cases of mitochondrial DNA depletion due to TK2 defect: An expanding spectrum. *Neurology*. 78(9): 644-8, 2012

Schoeler M, Loetscher PD, Rossaint R, Fahlenkamp AV, Eberhardt G, Rex S, **Weis J**, Coburn M. Dexmedetomidine is neuroprotective in an in vitro model for traumatic brain injury. *BMC Neurol*. 11: 12:20, 2012

Schwartz V, Krüttgen A, **Weis J**, Weber C, Ostendorf T, Lue H, Bernhagen J. Role for CD74 and CXCR4 in clathrin-dependent endocytosis of the cytokine MIF. *Eur J Cell Biol*. 91(6-7): 435-49, 2012

Saygili E, Kluttig R; Saygili Es, Rackauskas G, **Weis J**, Marx N, Schauerte P, Rana OR. Age-related regional differences in cardiac nerve growth factor expression. *Age (Dordr)*. 34(3): 659-67, 2012

Stratogianni A, Tosch M, Schlemmer H, **Weis J**, **Katona I**, Isenmann S, Haensch CA. Bortezomib-induced severe autonomic neuropathy. *Clin Auton Res*. 22(4): 199-202, 2012

Wan J, Yourshaw M, Mamsa H, Rudnik-Schöneborn S, Menezes MP, Hong JE, Leong DW, **Senderek J**, Salman MS, Chitayat D, Seeman P, von Moers A, Graul-Neumann L, Kornberg AJ, Castro-Gago M, Sobrido MJ, Sanefuji M, Shieh PB, Salamon N, Kim RC, Vinters HV, Chen Z, Zerres K, Ryan MM,

Nelson SF, Jen JC. Mutations in the RNA exosome component gene EXOSC3 cause pontocerebellar hypoplasia and spinal motor neuron degeneration. *Nature Genet.* 44(6): 704-8, 2012

Fischer C, Trajanoski S, Papić L, Windpassinger C, Bernert G, Freilinger M, Schabhüttl M, Arslan-Kirchner M, Javaher-Haghighi P, Plecko B, **Senderek J**, Rauscher C, Löscher WN, Pieber TR, Janecke AR, Auer-Grumbach M. SNP array-based whole genome homozygosity mapping as the first step to a molecular diagnosis in patients with Charcot-Marie-Tooth disease. *J Neurol.* 259(3): 515-23, 2012

Guergueltcheva V, Müller JS, Dusl M, **Senderek J**, Oldfors A, Lindbergh C, Maxwell S, Colomer J, Mallebrera CJ, Nascimento A, Vilchez JJ, Muelas N, Kirschner J, Nafissi S, Kariminejad A, Nilipour Y, Bozorgmehr B, Najmabadi H, Rodolico C, Sieb JP, Schlotter B, Schoser B, Herrmann R, Voit T, Steinlein OK, Najafi A, Urtizberea A, Soler DM, Muntoni F, Hanna MG, Chaouch A, Straub V, Bushby K, Palace J, Beeson D, Abicht A, Lochmüller H. Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. *J Neurol.* 259(5): 838-50, 2012

Beier D, Schriefer B, Brawanski K, Hau P, **Weis J**, Schulz JB, Beier CP. Efficacy of clinically relevant temozolomide dosing schemes in glioblastoma cancer stem cell lines. *J Neurooncol.* 109(1): 45-52, 2012

Fries M, Brücken A, Çizen A, Westerkamp M, Löwer C, Deike-Glindemann J, Schnorrenberger NK, Rex S, Coburn M, **Nolte KW**, **Weis J**, Rossaint R, Derwall M. Combining xenon and mild therapeutic hypothermia preserves neurological function after prolonged cardiac arrest in pigs. *Crit Care Med.* 40(4): 1297-1303, 2012

Galldiks N, Langen KJ, Holy R, Pinkawa M, Stoffels G, **Nolte KW**, Kaiser HJ, Filss CP, Fink GR, Coenen HH, Eble MJ, Piroth MD. Assessment of treatment response in patients with glioblastoma using O-(2-18F-fluoroethyl)-L-tyrosine PET in comparison to MRI. *J Nucl Med.* 53(7): 1048-57, 2012

van Neerven SG, Bozkurt A, O'Dey DM, Scheffel J, Boecker AH, Stromps JP, Dunda S, **Brook GA**, Pallua N. Retrograde tracing and toe spreading after experimental autologous nerve transplantation and crush injury of the sciatic nerve: a descriptive methodological study. *J Brachial Plex Peripher Nerve Inj.* 7(1): 5, 2012

Beier CP, Kumar P, Meyer K, Leukel P, Bruttel V, Aschenbrenner I, Riemenschneider MJ, Fragoulis A, Rümmele P, Lamszus K, Schulz JB, **Weis J**, Bogdahn U, Wischhusen J, Hau P, Spang R, Beier D. The cancer stem cell subtype determines immune infiltration of glioblastoma. *Stem Cells Dev.* 21(15): 2753-61, 2012

Creus KK, De Paepe B, **Weis J**, De Bleecker JL. The multifaceted character of lymphotoxin β in inflammatory myopathies and muscular dystrophies. *Neuromuscul Disord.* 22(8): 712-9, 2012

Claeys KG, Schrading S, Bozkurt A, Friedrich-Frekxa A, Pallua N, Kuhl C, Schulz JB, **Weis J**. Myopathy with lobulated fibers, cores, and rods caused by a mutation in collagen VI. *Neurology.* 79(23): 2288-90, 2012

Scholtes F, **Brook G**, Martin D. Spinal cord injury and its treatment: current management and experimental perspectives. *Adv. Tech. Stand. Neurosurg.* 38: 29-56, 2012

Hodde D, **Gerardo-Nava JL**, Deumens R, Mey J, **Brook GA**. Electrospinning of Nanofibres for Repair of the Injured Peripheral Nervous System. In: *Nanomedicine and the Nervous System*. Ed. Preedy VR. Science Publishers, New Hampshire USA. ISBN 9781578087280, 2012

Mey J, **Brook G**, **Hodde D**, Kriebel A. Electrospun Fibers as Substrates for Peripheral Nerve Regeneration. *Adv Polym Sci.* 246: 131-70, 2012

Publications 2011

Schröder JM, Klossok T, **Weis J**. Oculopharyngeal muscle dystrophy: fine structure and mRNA expression levels of PABPN1. *Clin Neuropathol.* 30(3): 94-103, 2011

Rana OR, Saygili E, Gemein C, Zink MD, Buhr A, Saygili E, Mischke K, **Nolte KW**, **Weis J**, Weber C, Marx N, Schauerte P. Chronic electrical neuronal stimulation increases cardiac parasympathetic tone by eliciting neurotrophic effects. *Circ Res.* 108(10): 1209-19, 2011

Saygili E, Pekassa M, Saygili E, Rackauskas G, Hommes D, Noor-Ebad F, Gemein C, Zink MD, Schwinger RH, **Weis J**, Marx N, Schauerte P, Rana OR. Mechanical stretch of sympathetic neurons induces VEGF expression via a NGF and CNTF signaling pathway. *Biochem Biophys Res Commun.* 410(1): 62-7, 2011

Weis J*, **Katona I***, Müller-Newen G, Sommer C, Necula G, Hendrich C, Ludolph AC, Sperfeld A-D. Small fiber neuropathy in ALS patients. *Neurology.* 76(23): 2024-9, 2011 *Equal contribution

Hanisch F, Müller T, Dietz A, Bitoun M, Kress W, **Weis J**, Stoltenburg G, Zierz S. Phenotype variability and histopathological findings in centronuclear myopathy due to DNM2 mutations. *J Neurol.* 258(6): 1085-90, 2011

Guelly C, Zhu PP, Leonardis L, Papic L, Zidar J, Schabhüttl M, Strohmaier H, **Weis J**, Strom TM, Baets J, Willems J, De Jonghe P, Reilly MM, Fröhlich E, Hatz M, Trajanoski S, Pieber TR, Janecke AR, Blackstone C, Auer-Grumbach M. Targeted High-Throughput Sequencing Identifies Mutations in atlastin-1 as a Cause of Hereditary Sensory Neuropathy Type I. *Am J Hum Genet.* 88(1): 99-105, 2011

Doorschodt BM, Schreinemachers MC, Behbahani M, Florquin S, **Weis J**, Staat M, Tolba RH. Hypothermic machine perfusion of kidney grafts: Which Pressure is Preferred? *Ann Biomed Eng.* 39(3): 1051-9, 2011

Saygili E, Schauerte P, Pekassa M, Saygilli E, Rackauskas G, Schwinger RH, **Weis J**, Weber C, Marx N, Rana OR. Sympathetic Neurons Express and Secrete MMP-2 and MT1-MMP to Control Nerve Sprouting via Pro-NFG Conversion. *Cell Mol Neurobiol.* 31(1): 17-25, 2011

Bozkurt A, Scheffel J, **Brook GA**, Joosten EA, Suschek CV, O'Dey DM, Pallua N, Deumens R. Aspects of static and dynamic motor function in peripheral nerve regeneration: SSI and CatWalk gait analysis. *Behav Brain Res.* 219(1): 55-62, 2011

Scholtes F, Theunissen E, Phan-Ba R, Adriaenssens P, **Brook G**, Franzen R, Gelan J, Schoenen J, Martin D. Post-mortem assessment of rat spinal cord injury and white matter sparing using inversion recovery-supported proton density magnetic resonance imaging. *Spinal Cord.* 49(3): 345-51, 2011

Bockelmann J, Klinkhammer K, von Holst A, Seiler N, Faissner A, **Brook GA**, Klee D, Mey J. Functionalization of electrospun poly(ϵ -caprolactone) fibers with the extracellular matrix-derived peptide GRGDS improves guidance of schwann cell migration and axonal growth. *Tissue Eng Part A.* 17(3-4): 475-86, 2011

Saporta MA, **Katona I**, Zhang X, Roper HP, McClelland L, Macdonald F, Brueton L, Blake J, Suter U, Reilly MM, Shy ME, Li J. Neuropathy in a human without the PMP22 gene. *Arch Neurol*. 68(6): 814-21, 2011

Hernandez-Lain A, Husson I, Monnier N, Farnoux C, Brochier G, Lacène E, Beuvin M, Viou M, Manéré L, **Claeys KG**, Fardeau M, Lunardi J, Voit T, Romero NB. De novo RYR1 heterozygous mutation (I4898T) causing lethal core-rod myopathy in twins. *Eur J Med Genet*. 54(1): 29-33, 2011

Katona I, Zhang X, Bai Y, Shy ME, Guo J, Yan Q, Hatfield J, Kupsky WJ, Li J. Distinct pathogenic processes between Fig4-deficient motor and sensory neurons. *Eur J Neurosci*. 33(8): 1401-10, 2011

Knosalla M, **Weis J**, Isenmann S, Haensch CA. L-Dihydroxyphenylserins as Therapy for the Rare Pure Autonomic Failure. *Klin Neurophysiol*. 42(2): 103-109, 2011

Bozkurt A, Dunda SE, Mon O'Dey D, **Brook GA**, Suschek CV, Pallua N. Epineurial sheath tube (EST) technique: an experimental peripheral nerve repair model. *Neurol Res*. 33(10): 1010-5, 2011

Bevilacqua JA, Monnier N, Bitoun M, Eymard B, Ferreira A, Monges S, Lubieniecki F, Taratuto AL, Laquerrière A, **Claeys KG**, Marty I, Fardeau M, Guicheney P, Lunardi J, Romero NB. Recessive RYR1 mutations cause unusual congenital myopathy with prominent nuclear internalization and large areas of myofibrillar disorganization. *Neuropathol Appl Neurobiol*. 37(3): 271-84, 2011

Oechtering J, Kirkpatrick PJ, Ludolph AG, Hans FJ, **Sellhaus B**, Spiegelberg A, Krings T. Magnetic microparticles for endovascular aneurysm treatment: in vitro and in vivo experimental results. *Neurosurgery*. 68(5): 1388-97, 2011

Menzel-Severing J, **Sellhaus B**, Laube T, Brockmann C, Bornfeld N, Walter P, Roessler G. Surgical Results and Microscopic Analysis of the Tissue Reaction following Implantation and Explantation of an Intraocular Implant for Epiretinal Stimulation in Minipigs. *Ophthalmic Res*. 46(4): 192-198, 2011

Bickenbach J, Biener I, Czaplik M, **Nolte K**, Dembinski R, Marx G, Rossaint R, Fries M. Neurological outcome after experimental lung injury. *Respir Physiol Neurobiol*. 179(2-3): 174-80, 2011

Bozkurt A, Boecker A, Van Neerven S, O'Dey DM, Opländer C, **Brook G**, Pallua N. A flexible, sterile, and cost effective retractor system for microsurgery. *Microsurgery* 31: 668-70, 2011

Publications 2010

Deumens R, Bozkurt A, Meek MF, Marcus M, Joosten E, **Weis J**, **Brook GA**. Repairing injured peripheral nerves: Bridging the gap. *Prog Neurobiol*. 92(3): 245-76, 2010

Nachreiner T, **Esser M**, **Tenten V**, Troost D, **Weis J**, **Krüttgen A**. Novel splice variants of the amyotrophic lateral sclerosis-associated gene VAPB expressed in human tissues. *Biochem Biophys Res Commun*. 394(3): 703-8, 2010

Sommer CL, Brandner S, Dyck PJ, Harati Y, LaCroix C, Lammens M, Magy L, Mellgren SI, Morbin M, Navarro C, Powell HC, Schenone AE, Tan E, Urtizbera A, **Weis J**. Peripheral Nerve Society Guideline on processing and evaluation of nerve biopsies. *J Peripher Nerv Syst*. 15(3): 164-75, 2010

Roehl AB, Hein M, Loetscher PD, Rossaint J, **Weis J**, Rossaint R, Coburn M. Neuroprotective properties of levosimendan in an in vitro model of traumatic brain injury. *BMC Neurol*. 10: 97, 2010

Bremer J, O'Connor T, Tiberi C, Rehrauer H, **Weis J**, Aguzzi A. Ablation of Dicer from murine Schwann cells increases their proliferation while blocking myelination. *PLoS One*. 5(8): e12450, 2010

Brücken A, Kaab AB, Kottmann K, Rossaint R, **Nolte KW**, **Weis J**, Fries M. Reducing the duration of 100% oxygen ventilation in the early reperfusion period after cardiopulmonary resuscitation decreases striatal brain damage. *Resuscitation*. 81(12): 1693-703, 2010

Urban PP, Wellach I, Faiss S, Layer P, Rosenkranz T, Knop K, **Weis J**. Subacute axonal neuropathy in Parkinson's disease with cobalamin and vitamin B6 deficiency under duodopa therapy. *Mov Disord*. 25(11): 1748-52, 2010

Claeys KG, Pellissier JF, Garcia-Bragado F, **Weis J**, Urtizberea A, Poza JJ, Cobo AM, Stoltenburg G, Figarella-Branger D, Willems PJ, Depuydt CE, Kleiner W, Pouget J, Piraud M, Brochier G, Romero NB, Fardeau M, Goebel HH, Bönnemann CG, Voit T, Eymard B, Laforet P. Myopathy with hexagonally cross-linked crystalloid inclusions: Delineation of a clinico-pathological entity. *Neuromuscul Disord*. 20(11): 701-8, 2010

Brauers E, **Dreier A**, Roos A, Wormland B, **Weis J***, **Krüttgen A***. Differential effects of myopathy-associated caveolin 3-mutants on growth factor signaling. *Am J Pathol*. 177(1): 261-70, 2010 *Equal contribution

Kaemmer D, Bozkurt A, Otto J, Junge K, Klinik C, **Weis J**, **Sellhaus B**, O'Dey DM, Pallua N, Jansen M, Schumpelick V, Klinge U. Evaluation of tissue components in the peripheral nervous system using Sirius red staining and immunohistochemistry: A comparative study (human, pig, rat). *J Neurosci Methods*. 190(1): 112-6, 2010

Rana OR, Schauerte P, Kluttig R, Schröder JW, Koenen RR, Weber C, **Nolte KW**, **Weis J**, Hoffmann R, Marx N, Saygili E. Acetylcholine as an age-dependent non-neuronal source in the heart. *Auton Neurosci*. 156(1-2): 82-9, 2010

Meyer C, Rana OR, Saygili E, Gemein C, Becker M, **Nolte K**, **Weis J**, Schimpf T, Knackstedt C, Mischke K, Hoffmann R, Kelm M, Pauza D, Schauerte P. Augmentation of Left Ventricular Contractility by Cardiac Sympathetic Neural Stimulation. *Circulation*. 121(11): 1286-94, 2010

Saygili E, Schauerte P, Küppers F, Heck L, **Weis J**, Weber C, Schwinger RH, Hoffmann R, Schröder JW, Marx N, Rana OR. Electrical stimulation of sympathetic neurons induces autocrine/paracrine effects of NGF mediated by TrkA. *J Mol Cell Cardiol*. 49(1): 79-87, 2010

Reilich P, Schramm N, Schoser B, Schneiderat P, Strigl-Pill N, Müller-Höcker J, Kress W, Ferbert A, Rudnik-Schöneborn S, Noth J, Lochmüller H, **Weis J**, Walter MC. Fascioscapulohumeral muscular dystrophy presenting with unusual phenotypes and atypical morphological features of vacuolar myopathy. *J Neurol*. 257(7): 1108-18, 2010

Flohr S, **Ewers P**, Fink GR, **Weis J**, **Krüttgen A**. Impaired Neurotrophin-3 signaling in a TrkAII mutant associated with hereditary polyneuropathy. *Exp Neurol*. 224(1): 318-20, 2010

Helfrich I, Scheffrahn I, Bartling S, **Weis J**, von Felbert V, Middleton M, Kato M, Ergün S, Schadendorf D. Resistance to antiangiogenic therapy is directed by vascular phenotype, vessel stabilization, and maturation in malignant melanoma. *J Exp Med*. 207(3): 491-503, 2010

Funke AD, **Esser M**, **Krüttgen A**, **Weis J**, Mitne-Neto M, Lazar M, Nishimura AL, Sperfeld AD, Trillenberg P, Senderek J, Krasnianski M, Zatz M, Zierz S, Deschauer M. The P56S mutation in the VAPB gene is not due to a single founder: the first European case. *Clin Genet*. 77(3): 302-3, 2010

Bremer J, Baumann F, Tiberi C, Wessig C, Fischer H, Schwarz P, Steele AD, Toyka KV, Nave KA, **Weis J**, Aguzzi A. Axonal prion protein is required for peripheral myelin maintenance. *Nature Neurosci.* 13(3): 310-18, 2010

Derwall M, Westerkamp M, Löwer C, Deike-Glindemann J, Schnorrenberger NK, Coburn M, **Nolte KW**, Gaisa N, **Weis J**, Siepmann K, Häusler M, Rossaint R, Fries M. Hydrogen sulfide does not increase resuscitability in a porcine model of prolonged cardiac arrest. *Shock.* 34(2): 190-5, 2010

Huttner HB, Richter G, Jünemann A, Kress W, **Weis J**, **Schröder JM**, Gal A, Doerfler A, Udd B, Schröder R. Incontinentia pigmenti-related myopathy or unsolved „double trouble“? *Neuromuscul Disord.* 20(2): 139-41, 2010

Evangelopoulos ME, Wüller S, **Weis J**, **Krüttgen A**. A role of nitric oxide in neurite outgrowth of neuroblastoma cells triggered by mevastatin or serum reduction. *Neurosci Lett.* 468(1): 28-33, 2010

Führmann T, **Gerardo-Nava J**, **Brook GA**. Tissue Engineering: Central Nervous System. In *Tissue Engineering*. Eds Pallua N, Suschek C. Springer Verlag, London, Heidelberg. 2010, in press

Deumens R, Bozkurt A, **Brook GA**. US Food and Drug Administration/Conformit Europe-approved absorbable nerve conduits for clinical repair of peripheral and cranial nerves. Commentary. *Ann Plast Surg.* 65(3): 371, 2010

Montzka K, **Führmann T**, Müller-Ehmsen J, Wöltje M, **Brook GA**. Growth factor and cytokine expression of human mesenchymal stromal cells is not altered in an in vitro model of tissue damage. *Cytotherapy.* 12(7): 870-80, 2010

Führmann T, **Hillen LM**, **Montzka K**, Wöltje M, **Brook GA**. Cell-cell interactions of human neural progenitor-derived astrocytes within a microstructured 3D-scaffold. *Biomaterials.* 31(30): 7705-15, 2010

Klinkhammer K, Bockelmann J, Simitzis C, **Brook GA**, Grafahrend D, Groll J, Möller M, Mey J, Klee D. Functionalization of electrospun fibers of poly(epsilon-caprolactone) with star shaped NCO-poly(ethylene glycol)-stat-poly(propylene glycol) for neuronal cell guidance. *J Mater Sci Mater Med.* 21(9): 2637-51, 2010

Führmann T, **Montzka K**, **Hillen LM**, **Hodde D**, **Dreier A**, Bozkurt A, Wöltje M, **Brook GA**. Axon growth-promoting properties of human bone marrow mesenchymal stromal cells. *Neurosci Lett.* 474(1): 37-41, 2010

Montzka K, **Führmann T**, Wöltje M, **Brook GA**. Expansion of human bone marrow-derived mesenchymal stromal cells: serum-reduced medium is better than conventional medium. *Cytotherapy.* 12(5): 587-92, 2010

Foret A, Quertainmont R, Botman O, Bouhy D, Amabili P, **Brook G**, Schoenen J, Franzen R. Stem cells in the adult rat spinal cord: plasticity after injury and treadmill training exercise. *J Neurochem.* 112(3): 762-72, 2010

van Neerven S, Joosten EAJ, **Brook GA**, Lambert CA, Mey J, **Weis J**, Marcus MA, Steinbusch HW, van Kleef M, Patijn J, Deumens R. Repetitive intrathecal VEGF 165 treatment has limited therapeutic effects after spinal cord injury in the rat. *J Neurotrauma.* 27(10): 1781-91, 2010

Mischke K, Zarse M, Schmid M, Gemein C, Brauers N, Spillner J, Dohmen G, Rana O, Saygili E, Knackstedt C, **Weis J**, Pauza D, Bianchi S, Schauerte P. Chronic augmentation of the parasympathetic

tone to the atrioventricular node: A nonthoracotomy neurostimulation technique for ventricular rate control during atrial fibrillation. *J Cardiovasc Electrophysiol.* 21(2): 193-9, 2010

Reinges MH, Krings T, Drexler AY, Ludolph A, **Sellhaus B**, Bovi M, Geibprasert S, Agid R, Scherer K, Hans FJ. Bare, bio-active and hydrogel-coated coils for endovascular treatment of experimentally induced aneurysms. Long-term histological and scanning electron microscopy results. *Interv Neuroradiol.* 16(2): 139-50, 2010

Geibprasert S, Krings T, Apitzsch J, Reinges MH, **Nolte KW**, Hans FJ. Subarachnoid hemorrhage following posterior spinal artery aneurysm. A case report and review of the literature. *Interv Neuroradiol.* 16(2): 183-90, 2010

Schilling S, Klotz P, **Weis J**, Gold R. Steroid responsive dementia syndrome and vasculitic polyneuritis. *Akt Neurologie.* 37(2): 80-2, 2010

Publications 2009

Loetscher PD, Rossaint J, Rossaint R, **Weis J**, Fries M, Fahlenkamp A, Ryang YM, Grottke O, Coburn M. Argon: neuroprotection in in vitro models of cerebral ischemia and traumatic brain injury. *Crit Care.* 13(6): R206, 2009

Weis J, **Nolte K**, Mader H, **Schröder JM**, Grehl H, Rada A, Zerres K, Senderek J. Late sporadic CMT4C-A new KIAA1985 mutation. *Expert Consult: Companion to Peripheral Neuropathy.* 171-173, 2009

Oertel M, **Nolte K**, Blaum M, **Weis J**, Gilsbach J, Korinth M. Primary intraventricular schwannomas. *Clin Neurol Neurosurg.* 111(9): 768-73, 2009

Schaakxs D, Bahm J, **Sellhaus B**, **Weis J**. Clinical and neuropathological study about the neurotization of the suprascapular nerve in obstetric brachial plexus lesions. *J Brachial Plex Peripher Nerve Inj.* 4: 15, 2009

Arnaud E, Zenker J, de Preux Charles AS, Stendel C, Roos A, Médard JJ, Tricaus N, **Weis J**, Suter U, Senderek J, Chrast R. SH3TC2/ KIAA1985 protein is required for proper myelination and the integrity of the node of Ranvier in the peripheral nervous system. *Proc Natl Acad Sci USA*106. (41): 17528-33, 2009

Moises T, Wüller S, Saxena S, Senderek J, **Weis J**, **Krüttgen A**. Proteasomal inhibition alters the trafficking of the neurotrophin receptor TrkA. *Biochem Biophys Res Commun.* 387(2): 360-4, 2009

Weis J, **Nikolin S**, **Nolte K**. Neurogenic muscular atrophy and selective fibre type atrophies: Crucial findings in the biopsy diagnosis of neuromuscular disease. *Pathologe.* 30(5): 379-83, 2009

Weis J. Myopathology: an update. *Pathologe.* 30(5):343-4, 2009

Weis J, **Nolte K**. Inflammatory and other myopathies and skeletal muscle vasculitis: The role of muscle and nerve biopsy. *Z Rheumatol.* 68(6): 459-64, 2009

Bergmann M, **Weis J**, Probst-Cousin S. Muscle biopsy: Indications and techniques. *Pathologe.* 30(5): 348-51, 2009

Weis J, **Nikolin S**, **Nolte K**. Muskel- und Nervenbiopsien: Aktuelle Aspekte. *Nervenheilkunde.* 9: 624-

26, 2009

Senderek J, Garvey SM, Krieger M, Guergueltcheva V, Urtizberea A, Roos A, Elbracht M, Stendel C, Tournev I, Mihailova V, Feit H, Tramonte J, Hedera P, Crooks K, Bergmann C, Rudnik-Schöneborn S, Zerres K, Lochmüller H, Seboun E, **Weis J**, Beckmann JS, Hauser MA, Jackson CE. Autosomal-dominant distal myopathy associated with a recurrent missense mutation in the gene encoding the nuclear matrix protein, matrin 3. *Am J Hum Genet.* 84: 511-18, 2009

Schröder JM. "Necklace" fibers as a late clue to the interpretation of the forgotten "trilaminar" fibers. *Acta Neuropathol.* 118: 317-8, 2009

Roessler GF, Laube T, Brockmann C, Kirschkamp T, Mazinani BA, Goertz M, Koch C, Kirsch I, **Sellhaus B**, Trieu HK, **Weis J**, Bornfeld N, Roethgen H, Messner A, Mokwa W, Walter P. Implantation and explantation of a wireless epiretinal retina implant device: Observations during the EPIRET 3 prospective clinical trial. *Invest Ophthalmol Vis Sci.* 50(6): 3003-08, 2009

Rossaint J, Rossaint R, **Weis J**, Fries M, Rex S, Coburn M. Propofol: neuroprotection in an in vitro model of traumatic brain injury. *Critical Care.* 13(2): R61, 2009

Weis J. Stellenwert der Muskelbiopsie in der Diagnostik der Muskelerkrankungen. *NeuroAktuell.* 5: 21-25, 2009

Bahm J, Ocampo-Pavez C, Disselhorst-Klug C, **Sellhaus B**, **Weis J.** Obstetric brachial plexus palsy. *Deutsches Ärzteblatt.* 6: 83-90, 2009

Evangelopoulos ME, **Weis J**, **Krüttgen A.** Mevastatin-induced neurite outgrowth of neuroblastoma cells via activation of EGFR. *J Neuroscience Res.* 87: 2138-44, 2009

Bozkurt A, Deumens R, Beckmann C, Olde Damink L, Schügner F, Heschel I, **Sellhaus B**, **Weis J**, Jahnen-Dechent W, **Brook G**, Pallua N. In vitro cell alignment obtained with a Schwann cell enriched microstructured nerve guide with longitudinal guidance channels. *Biomaterials.* 30(2): 169-79, 2009

Koopmans G, Deumens R, Buss A, Geoghegan L, Mu Myint A, Honig W, Kern N, Joosten E, Noth J, **Brook G.** Acute rolipram/thalidomide treatment improves tissue sparing and locomotion after experimental spinal cord injury. *Exp Neurol.* 216(2): 490-98, 2009

Derwall M, Coburn M, Rex S, Hein M, Rossaint R, Fries M. Xenon: recent developments and future perspectives. *Minerva Anestesiol.* 75(1-2): 37-45, 2009

Rana OR, Saygili E, Meyer C, Gemein C, **Krüttgen A**, Andrzejewski MG, Ludwig A, Schotten U, Schwinger RH, Weber C, **Weis J**, Mischke K, Rassaf T, Kelm M, Schauerte P. Regulation of nerve growth factor in the heart: the role of the calcineurin- NFAT pathway. *J Mol Cell Cardiol.* 46(4): 568- 78, 2009

Saygili E, Rana OR, Meyer C, Gemein C, Andrzejewski MG, Ludwig A, Weber C, Schotten U, **Krüttgen A**, **Weis J**, Schwinger RH, Mischke K, Rassaf T, Kelm M, Schauerte P. The angiotensin-calcineurin-NFAT pathway mediates stretch-induced up-regulation of matrix metalloproteinases-2/-9 in atrial myocytes. *Basic Res Cardiol.* 104(4): 435-448, 2009

Creus K, De Paepe B, Werbruggen B, Vervaeke V, **Weis J**, De Bleecker J. Distribution of the NF- κ B complex in the inflammatory exudates characterizing the idiopathic inflammatory myopathies. *Ann NY Acad Sci.* 1173: 370-7, 2009

De Paepe B, Creus K, Martin J, **Weis J**, De Bleecker J. A dual role for HSP90 and HSP70 in the inflammatory neuropathies: from muscle fiber protection to active invasion by macrophages. *Ann NY Acad Sci.* 1173: 463-9, 2009

Montzka K, Lassonczyk N, Tschöke B, Neuss S, **Führmann T**, Franzen R, Smeets R, **Brook G**, Wöltje M. Neural differentiation potential of human bone marrow-derived mesenchymal stromal cells: misleading marker gene expression. *BMC Neuroscience.* 10:16, 2009

Möllers S, Heschel I, Olde Damink L, Schügner F, Deumens R, Müller B, Bozkurt A, **Nava J**, Noth J, **Brook G**. Cytocompatibility of a novel, longitudinally microstructured collagen scaffold intended for nerve tissue repair. *Tissue Eng Part A.* 15(3): 461-72, 2009

Gerado-Nava J, Führmann T, Klinkhammer K, Seiler N, Mey J, Klee D, Möller M, Dalton P, **Brook G**. Human neural cell interactions with orientated electrospun nanofibers in vitro. *Nanomedicine.* 4(1): 11-30, 2009

Klinkhammer K, Seiler N, Grafahrend D, **Gerardo-Nava JL**, Mey J, **Brook G**, Möller M, Dalton PD, Klee D. Deposition of electrospun fibers on reactive substrates for in vitro investigations. *Tissue Eng Part C Methods.* 15(1): 77-85, 2009

Bozkurt A, Deumens R, Beckmann C, Olde Damink L, Schügner F, Heschel I, **Sellhaus B**, **Weis J**, Jahnen-Dechent W, **Brook GA**, Pallua N. In vitro cell alignment obtained with a Schwann cell enriched microstructured nerve guide with longitudinal guidance channels. *Biomaterials.* 30(2): 169-79, 2009

Andres RH, Guzman R, **Weis J**, Brekenfeld C, Fandino J, Seiler RW. Lhermitte-Duclos disease with atypical vascularization--case report and review of the literature. *Clin Neuropathol.* 28(2): 83-90, 2009

Fries M, Coburn M, **Nolte KW**, Timper A, Kottmann K, Kuru TH, **Weis J**, Rossaint R. Early administration of xenon or isoflurane may not improve functional outcome and cerebral alterations in a porcine model of cardiac arrest. *Resuscitation.* 80(5): 584-90, 2009

Balciuniene N, Tamasauskas A, Valanciute A, Deltuva V, Vaitiekaitis G, Gudaviciene I, **Weis J**, von Keyserlingk DG. Histology of human glioblastoma transplanted on chicken chorioallantoic membrane. *Medicina (Kaunas).* 45(2): 123-31, 2009

Buss A, Pech K, Kakulas BA, Didier M, Schoenen J, Noth J, **Brook G**. NG2 and phosphacan are present in the astroglial scar after human traumatic spinal cord injury. *BMC Neurology.* 9: 32, 2009

Rotthier A, Baets J, De Vriendt E, Jacobs A, Auer-Grumbach M, Le'vy N, Bonello-Palot N, Sebnem Kilic S, **Weis J**, Nascimento A, Swinkels M, Kruyt MC, Jordanova A, De Jonghe P, Timmerman V. Genes for hereditary sensory and autonomic neuropathies: a genotype-phenotype correlation. *Brain.* 132: 2699-711, 2009

Schroeder A, Ertl-Wagner B, Britsch S, **Schröder J**, **Nikolin S**, **Weis J**, Müller-Felber W, Koerte I, Stehr M, Berweck S, Borggraefe I, Heinen F. Muscle biopsy substantiates long-term MRI alterations one year after a single dose of botulinum toxin injected into the lateral gastrocnemius muscle of healthy volunteers. *Mov Disord.* 24(10): 1494-503, 2009

Publications 2008

Weis J. Twenty-five years of the Neuromuscular Disease reference Center of the German Society for Neuroanatomy. *Nervenarzt*. 79: 958-60, 2008

Sommer C, Brandner S, Dyck PJ, Magy L, Mellgren SI, Morbin M, Schenone A, Tan E, **Weis J.** 147th ENMC international workshop: guideline on processing and evaluation of sural nerve biopsies, 15-17 December 2006, Naarden, The Netherlands. *Neuromuscul Disord*. 18: 90-96, 2008

Koy A, Ilkovski B, Laing N, North K, **Weis J**, Neuen-Jacob E, Maytepek E. Nemaline myopathy with exclusively intranuclear rods and a novel mutation in ACTA1 (Q139H). *Neuropediatrics*. 39: 1-5, 2008

Fries M, **Nolte K**, Demir F, Kottmann K, Timper A, Coburn M, **Weis J**, Rossaint R. Neurocognitive performance after cardiopulmonary resuscitation in pigs. *Critical Care Med*. 36: 842-847, 2008

Nolte KW, Hans VJ, Schattenfroh C, **Weis J**, **Schröder JM**. Perineurial cells filled with collagen in „atypical“ Cogan's syndrome. *Acta Neuropathol*. 115: 589-96, 2008

Urban PP, Kaczmarek E, Wellach I, Brüning R, Brüllke N, Schulte C, Knop K, **Weis J.** [Neurolymphomatosis : Subacute sensorimotor polyneuropathy as a first sign of non-Hodgkin's B cell lymphoma.] *Nervenarzt*. 79: 699-702, 2008

Guzman R, Altrichter S, El-Koussy M, Gralla J, **Weis J**, Barth A, Seiler RW, Schroth G, Lövblad KO. Contribution of the apparent diffusion coefficient in perilesional edema for the assessment of brain tumors. *J Neuroradiol*. 35(4): 224-9, 2008

Fries M, **Nolte K**, Timper A, Kottmann K, **Weis J**, Rossaint R. Xenon reduces neurohistopathological damage and improves the early neurological deficit after cardiac arrest in pigs. *Critical Care Med*. 36: 2420-26, 2008

Bozkurt A, Tholl S, Wehner S, Tank J, Cortese M, O'Dey D, Deumens R, Lassner F, Schügner F, Gröger A, Smeets R, **Brook G**, Pallua N. Evaluation of functional nerve recovery with Visual-SSI--a novel computerized approach for the assessment of the static sciatic index (SSI). *J Neurosci Methods*. 170(1): 117-122, 2008

Bozkurt A, Deumens R, Scheffel J, O'Dey DM, **Weis J**, Joosten EA, **Führmann T**, **Brook G**, Pallua N. CatWalk gait analysis in assessment of functional recovery after sciatic nerve injury. *J Neurosci Methods*. 173(1): 91-98, 2008

Weis J. Stellenwert der Muskelbiopsie in der Diagnostik der Muskelerkrankungen. *NeuroAktuell*. 501-03, 2008

Scholtes F, Phan-Ba R, Theunissen E, Adriaensens P, **Brook G**, Franzen R, Bouhy D, Gelan J, Martin D, Schoenen J. Rapid, postmortem 9.4T MRI of spinal cord injury: Correlation with histology and survival times. *J Neurosci Methods*. 174(2): 157-67, 2008

Buss A, Pech K, Kakulas BA, Martin D, Schoenen J, Noth J, **Brook G**. TGF-beta1 and TGF-beta2 expressions after traumatic human spinal cord injury. *Spinal Cord*. 46(5): 364-71, 2008

Dafotakis M, Sparing R, Eickhoff SB, Boy C, **Nikolin S**, Fink GR. Postinfectious focal necrotizing myopathy. *Clin Nucl Med*. 33(7): 500-1, 2008

Nolte KW, Janecke AR, Vorgerd M, **Weis J**, **Schröder JM**. Congenital type IV glycogenosis: the spectrum of pleomorphic polyglucosan bodies in muscle, nerve, and spinal cord with two novel mutations in the GBE1 gene. *Acta Neuropathol*. 116(5): 491-506, 2008

Rudnik-Schöneborn S, **Weis J**, Kress W, Häusler M, Zerres K. Becker's muscular dystrophy aggravating facioscapulohumeral muscular dystrophy - double trouble as an explanation for an atypical phenotype. *Neuromuscul Disord.* 18(11): 881-5, 2008

Jeub M, Bitoun M, Guicheney P, Kappes-Horn K, Strach K, Druschky KF, **Weis J**, Fischer D: Dynamitin 2 related centronuclear myopathy: Clinical, histological and genetic aspects of further patients and review of the literature. *Clin Neuropathol.* 27: 430-438, 2008

Roessler G, Laube T, Brockmann C, Kirschkamp T, Mazinani B, Goertz M, Koch C, Kirsch I, **Sellhaus B**, Trieu HK, **Weis J**, Bornfeld N, Röthgen H, Messner A, Mokwa W, Walter P. Implantation and explantation of a wireless epiretinal retina implant device in blind RP patients. *Invest Ophthalmol Vis Sci.* 50(6): 3003-8, 2008

Schröder J.M.: Congenital Fiber Type Disproportion In: Lang F. (Hrsg.) *Encyclopedia of Molecular Mechanisms of Disease* ISBN 978-3-540-33445-3, Springer Medizinverlag 2008 (2nd Edition)

Schröder J.M.: Ferritinopathy In: Lang F. (Hrsg.) *Encyclopedia of Molecular Mechanisms of Disease* ISBN 978-3-540-33445-3, Springer Medizinverlag 2008 (2nd Edition)

Publications 2007

Ponzoni M, Berger F, Chassagne-Clement C, Tinguely M, Jouvett A, Ferreri AJ, Dell'oro S, Terreni MR, Doglioni C, **Weis J**, Cerati M, Milani M, Iuzzolino P, Motta T, Carbone A, Pedrinis E, Sanchez J, Blay JY, Reni M, Conconi A, Bertoni F, Zucca E, Cavalli F, Borisch B; on Behalf of the International Extranodal Lymphoma Study Group (IELSG). Reactive perivascular T-cell infiltrate predicts survival in primary central nervous system B-cell lymphomas. *Br J Haematol.* 138(3): 316-23, 2007

Stendel C, Roos A, Deconinck T, Pereira J, Castagner F, Niemann A, Kirschner J, Korinthenberg R, Ketelsen UP, Battaloglu E, Parman Y, Nicholson G, Ouvrier R, Seeger J, Jonghe PD, **Weis J**, **Krüttgen A**, Rudnik-Schöneborn S, Bergmann C, Suter U, Zerres K, Timmerman V, Relvas JB, Senderek J. Peripheral Nerve Demyelination Caused by a Mutant Rho GTPase Guanine Nucleotide Exchange Factor, Frabin/FGD4. *Am J Hum Genet.* 81(1): 158-64, 2007

Buss A, Pech K, Kakulas BA, Martin D, Schoenen J, Noth J, **Brook GA**. Matrix metalloproteinases and their inhibitors in human traumatic spinal cord injury. *BMC Neurology.* 7: 17, 2007

Wühl E, Kogan J, Zurowska A, Matejas V, Vandevoorde RG, Aigner T, Wendler O, Lesniewska I, Bouverier R, Reis A, **Weis J**, Cochat P, Zenker M. Neurodevelopmental deficits in Pierson (microcoria-congenital nephrosis) syndrome. *Am J Med Genet.* A143A: 311-19, 2007

Thiex R, **Weis J**, Krings T, Barreiro S, Yakisikli-Alemi F, Gilsbach JM, Rohde V. Addition of intravenous N-methyl-D-aspartate receptor antagonists to local fibrinolytic therapy for the optimal treatment of experimental intracerebral hemorrhages. *J Neurosurg.* 106: 314-20, 2007

Smilowitz HM, Weissenberger J, **Weis J**, Brown JD, O'Neill RJ, Laissue JA. Orthotopic transplantation of v-src-expressing glioma cell lines into immunocompetent mice: establishment of a new transplantable in vivo model for malignant glioma. *J Neurosurg.* 106: 652-59, 2007

Schoser BGH, **Schröder JM**, Grimm T, Sternberg D., Kress W. A large german kindred with cold-aggravated myotonia and a heterozygous A1481D mutation in the *SCN4A* gene. *Muscle Nerve.* 35: 599-606, 2007

Yen K, Lovblad KO, Scheurer E, Ozdoba C, Thali MJ, Aghayev E, Jackowski C, Anon J, Frickey N, Zwygart K, **Weis J**, Dirnhofer R. Post-mortem forensic neuroimaging: Correlation of MSCT and MRI findings with autopsy results. *Forensic Sci Int* 173: 21-35, 2007

Johann V, Schiefer J, Sass C, Mey J, Brook G, **Krüttgen A**, Schlangen C, Benreuther C, Schachner M, Dihne M, Kosinski CM. Time of transplantation and cell preparation determine neural stem cell survival in a mouse model of Huntington's disease. *Exp Brain Res*. 177(4): 458-70, 2007

Ramaekers VT, **Weis J**, Sequeira JM, Quadros EV, Blau N. Mitochondrial Complex I Encephalomyopathy and Cerebral 5-Methyltetrahydrofolate Deficiency. *Neuropediatrics*. 38: 1-4, 2007

Ueçeyler N, **Sellhaus B**, Sommer C, Reiners K. Polyneuropathien bei Porphyrie. *Nervenheilkunde*. 26: 763-68, 2007

Deumens R, **Lübberts M**, Jaken RJP, Meijs MFL, Thurlings RM, Honig WMM, Schachner M, **Brook GA**, Joosten EAJ. Mice lacking L1 have reduced CGRP fibre in-growth into spinal transection lesions. *Neuroscience Letters*. 420: 277-81, 2007

Fathi AR, Vassella E, Arnold M, Curschmann J, Reinert M, Vajtai I, **Weis J**, Deiana G, Mariani L. Objective response to radiation therapy and long-term survival of patients with WHO grade II astrocytic gliomas with known LOH 1p/19q status. *Strahlenther Onkol*. 183(9): 517-22, 2007

Andres RH, Guzman R, **Weis J**, Schroth G, Barth A. Granuloma formation and occlusion of an unruptured aneurysm after wrapping. *Acta Neurochir*. 149(9): 953-58, 2007

Bronfman FC, Escudero CA, **Weis J**, **Krüttgen A**. Endosomal transport of neurotrophins: roles in signaling and neurodegenerative diseases. *Dev Neurobiol*. 67(9): 1183-203, 2007

Häusler M, **Sellhaus B**, Scheithauer S, Gaida B, Kuropka S, Siepmann K, Panek A, Berg W, Teubner A, Ritter K, Kleines M. Myocarditis in newborn wild-type BALB/c mice infected with the murine gamma herpesvirus MHV-68. *Cardiovascular Research*. 76: 323-30, 2007

Moises T, Dreier A, Flohr S, Esser M, Brauers E, Reiss K, Merken D, Weis J, Krüttgen A. Tracking TrkA's Trafficking: NGF Receptor Trafficking Controls NGF Receptor Signaling. *Mol Neurobiol*. 35(2): 151-9, 2007

Hehr U, Bauer P, Winner B, Schuele R, Olmez A, Koehler W, Uyanik G, Engel A, Lenz D, Seibel A, Hehr A, Ploetz S, Gamez J, Rolfs A, **Weis J**, Ringer TM, Bonin M, Schuierer G, Marienhagen J, Bogdahn U, Weber BHF, Topaloglu H, Schoels L, Riess O, Winkler J. Long-term course and mutational spectrum of spatacsin-linked spastic paraplegia. *Ann Neurol*. 62: 656-65, 2007

Bozkurt A, **Brook GA**, Moellers S, Lassner F, **Sellhaus B**, **Weis J**, Woeltje M, Tank J, Beckmann C, Fuchs P, Olde Daminik L, Schügner F, Heschel I, Pallua N. In vitro assessment of axonal growth using dorsal root ganglia explants in a novel three-dimensional collagen matrix. *Tiss Eng*. 13(12): 2971-79, 2007

Buss A, Pech K, Kakulas BA, Martin D, Schoenen J, Noth J, **Brook GA**. Growth-modulating molecules are associated with invading Schwann cells and not astrocytes in human traumatic spinal cord injury. *Brain*. 130: 940-53, 2007

Kosinski CM, Schlangen C, Gellerich FN, Gizatullina Z, Deschauer M, Schiefer J, Young AB, Landwehrmeyer GB, Toyka KV, **Sellhaus B**, Lindenberg KS. Myopathy as a first symptom of Huntington's disease in a Marathon runner. *Mov Disord.* 22: 1637-40, 2007

Wirths O, **Weis J**, Kaye R, Saido TC, Bayer TA. Age-dependent axonal degeneration in an Alzheimer mouse model. *Neurobiol Aging* 28: 1689-99, 2007

Koopmans GC, Deumens R, **Brook G**, Gerver J, Honig WM, Hamers FP, Joosten EA. Strain and locomotor speed affect over-ground locomotion in intact rats. *Physiol Behav.* 92: 993-1001, 2007

Wuhl E, Kogan J, Zurowska A, Matejas V, Vandevoorde RG, Aigner T, Wendler O, Lesniewska I, Bouvier R, Reis A, **Weis J**, Cochat P, Zenker M. Neurodevelopmental deficits in Prierson (microcoria-congenital nephrosis) syndrom. *Am J Med Genet A.* 143(4): 311-319, 2007

Publications 2006

Fries M, Weis J, Rossaint R. Is Xenon really neuroprotective after cardiac arrest? *Anesthesiology.* 104: 211, 2006

Yen K, Lövblad KO, Scheurer E, Ozdoba C, Thali MJ, Aghayev E, Jackowski C, Anon J, Frickey N, **Weis J**, Zwygart K, Bratzke H, Dirnhofer R. Line scan diffusion tensor imaging of the post-traumatic brainstem: Changes with neuropathological correlation. *AJNR Am J Neuroradiol.* 27: 70-3, 2006

Wirths O, **Weis J**, Szczygielski J, Multhaup G, Bayer TA. Axonopathy in an APP/PS1 transgenic mouse model of Alzheimer's disease. *Acta Neuropathol.* 111: 312-19, 2006

Ramelli G, Joncourt F, Luetsch J, **Weis J**, Tolnay M, Burgunder J. Becker muscular dystrophy with marked divergence between clinical and molecular genetic findings: case series. *Swiss Med Wkly.* 136: 189-93, 2006

Möller JC, **Krüttgen A**, Burmester R, **Weis J**, Oertel WH, Shooter EM. Release of interleukin-6 via the regulated secretory pathway in PC12 cells. *Neurosci Lett.* 400: 75-9, 2006

Landolt HP, Glatzel M, Blättler Th, Achermann P, Roth C, Mathis J, **Weis J**, Tobler I, Aguzzi A, Bassetti CL. Sleep-wake disturbances in sporadic Creutzfeldt-Jakob Disease. *Neurology.* 66: 1418-24, 2006

Hergersberg M, Mariani L, Vassella E, Murtin C, **Weis J**, Moschopoulos M, Laeng H, Landolt H, Huber A, Roelcke U. Age at diagnosis and loss of heterozygosity on chromosome 1p and 19 q in oligodendroglial tumors. *J NeuroOncol.* 80:215-17, 2006

Verhoeven K, Claeys KG, Züchner S, **Schröder JM, Weis J**, Ceuterick C, Jordanova A, Nelis E, De Vriendt E, Van Hul M, Seeman P, Mazanec R, Saifi GM, Sziget K, Mancias P, Butler IJ, Kochanski A, Ryniewicz B, De Bleecker J, Van den Bergh P, Verellen C, Van Coster R, Goeman N, Robberecht W, Rasic VM, Nevo Y, Tournier I, Guergueltcheva V, Roelens F, Vieregge P, Vinci P, Moreno MT, Christen HJ, Shy ME, Lupski JR, Vance JM, De Jonghe P, Timmerman V. MFN2 mutation distribution and genotype/phenotype correlation in Charcot-Marie-Tooth type 2. *Brain.* 129: 2093-102, 2006

Mariani L, Deiana G, Vassella E, Fathi AR, Murtin C, Arnold M, Vajtai I, **Weis J**, Siegenthaler P, Schoesberger M, Reinert MM. Loss of heterozygosity 1p36 and 19q13 is a prognostic factor for overall survival in patients with diffuse WHO grade 2 gliomas treated without chemotherapy. *J Clin Oncol.* 24: 4758-63, 2006

Ramelli G. P., Gallati S., **Weis J.**, Krähenbühl S., Burgunder J.-M. Point Mutation tRNA^{Ser(UCN)} in a Child With Hearing Loss and Myoclonus Epilepsy. *J Child Neurol.* 21: 253-55, 2006

Wirths O, **Weis J**, Kayed R, Saido TC, Bayer TA. Age-dependent axonal degeneration in an Alzheimer mouse model. *Neurobiol Aging.* 28(11): 1689-99, 2006

Krings T, Busch C, **Sellhaus B**, Drexler AY, Bovi M, Hermanns-Sachweh B, Scherer K, Gilsbach JM, Thron A, Hans FJ. Long-term histological and scanning electron microscopy results of endovascular and operative treatments of experimentally induced aneurysms in the rabbit. *Neurosurgery.* 400(1-2): 911-23, 2006

Schröder JM: Neuropathology of Charcot-Marie-Tooth and Related Disorders *NeuroMolecular Medicine.* 8: 23-42, 2006

Krüttgen A, Schneider I, Weis J. The dark side of the NGF family: Neurotrophins in neoplasias. *Brain Pathol.* 16: 304-10, 2006

Publications 2005

Yen K, Sonnenschein M, Thali MJ, Ozdoba C, **Weis J**, Zwygart K, Aghayev E, Jackowski C, Dirnhofer R. Postmortem multislice computed tomography and magnetic resonance imaging of odontoid fractures, atlantoaxial distractions and ascending medullary edema. *Int J Legal Med.* 119: 129-36, 2005

Walther LE, Ilgner J, Oehme A, Schmidt P, **Sellhaus B**, Gudziol H, Beleites E, Westhofen M. Infectious mononucleosis. *HNO.* 53: 383-94, 2005

von Felbert V, Córdoba F, Weissenberger J, Vallan C, Kato M, Nakashima I, Braathen LR, **Weis J.** Interleukin-6 gene ablation in a transgenic mouse model of spontaneous malignant skin melanoma. *Am J Pathol.* 166: 831-41, 2005

Thiex R, Hans FJ, Krings T, **Sellhaus B**, Gilsbach JM. Technical pitfalls in a porcine brain retraction model. The impact of brain spatula on the retracted brain tissue in a porcine model: a feasibility study and its technical pitfalls. *Neuroradiology.* 47: 765-73, 2005

Szurman P, Warga M, Roters S, Grisanti S, Heimann U, Aisenbrey S, Rohrbach JM, **Sellhaus B**, Ziemssen F, Bartz-Schmidt KU. Experimental implantation and long-term testing of an intraocular vision aid in rabbits. *Arch Ophthalmol.* 123: 964-69, 2005

Schröder JM. Ferritinopathy: diagnosis by muscle or nerve biopsy, with a note on other nuclear inclusion body diseases. *Acta Neuropathol (Berl).* 109: 109-14, 2005

Saxena S, Howe CL, Cosgaya JM, Steiner P, Hirling H, Chan JR, **Weis J, Krüttgen A.** Differential endocytic sorting of p75NTR and TrkA in response to NGF: a role for the ubiquitin/proteasome system in trafficking of TrkA into multivesicular bodies. *Mol Cell Neurosci.* 28: 571-87, 2005

Ozdoba C, **Weis J**, Plattner T, Dirnhofer R, Yen K. Fatal scuba diving incident with massive gas embolism in cerebral and spinal arteries. *Neuroradiology.* 47: 411-16, 2005

Mawrin C, Schneider T, Firsching R, Wiedemann FR, Dietzmann K, Bornemann A, Romeike BF, **Sellhaus B**, von Deimling A. Assessment of tumor cell invasion factors in gliomatosis cerebri. *J Neurooncol.* 73: 109-15, 2005

Mawrin C, Kirches E, Schneider-Stock R, Boltze C, Vorwerk CK, Mawrin A, von Deimling A, Stoltenburg-Didinger G, Bornemann A, Romeike B, **Sellhaus B**, Dietzmann K. Alterations of cell cycle regulators in gliomatosis cerebri. *J Neurooncol.* 72: 115-22, 2005

Marcao AM, Wiest R, Schindler K, Wiesmann U, **Weis J**, Schroth G, Miranda MSC, Sturzenegger M, Gieselmann V. Adult onset metachromatic leukodystrophy without electroclinical peripheral nervous system involvement: a new mutation in the ARSA gene. *Arch Neurol.* 62: 309-13, 2005

Loeffler S, Fayard B, **Weis J**, Weissenberger J. Interleukin-6 induces vascular endothelial growth factor (VEGF) expression in the mouse brain: evidence that IL-6 induces VEGF promoter activity via STAT3 and Sp1. *Int J Cancer.* 115: 202-13, 2005

Kaindl AM, Jakubiczka S, Luecke T, Bartsch O, **Weis J**, Stoltenburg-Didinger G, Aksu F, Oexle K, Koehler K, Huebner A. A homozygous microdeletion of chromosome 4q11-q12 causes severe limb girdle muscular dystrophy type 2E with joint hyperlaxity and contractures. *Hum Mutat.* 26: 279-80, 2005

Jenne DE, Kley RA, Vorgerd M, **Schröder JM**, **Weis J**, Reimann H, Albrecht B, Nürnberg P, Thiele H, Müller CR, Meng G, Witt CC, Labeit S. Limb girdle muscular dystrophy in a sibling pair with a homozygous Ser606Leu mutation in the alternatively spliced IS2 region of calpain 3. *Biol Chem.* 386: 61-7, 2005

Hoelzinger DB, Mariani L, **Weis J**, Woyke T, Berens TJ, McDonough WS, Sloan A, Coons SW, Berens ME. Gene expression profile of glioblastoma multiforme invasive phenotype points to new therapeutic targets. *Neoplasia.* 7: 7-16, 2005

Häusler M, **Sellhaus B**, Scheithauer S, Engler M, Alberg E, Teubner A, Ritter K, Kleines M: Murine gammaherpesvirus-68 infection of mice: A new model for human cerebral Epstein-Barr virus infection. *Ann Neurol.* 57: 600-3, 2005

Fries M, Bickenbach J, Henzler D, Beckers S, Dembinski R, **Sellhaus B**, Rossaint R, Kuhlen R. S-100 protein and neurohistopathologic changes in a porcine model of acute lung injury. *Anesthesiology.* 102: 761-7, 2005

Fayard B, Loeffler S, **Weis J**, Vogelin E, **Krüttgen A**. The secreted brain-derived neurotrophic factor precursor pro-BDNF binds to TrkB and p75NTR but not to TrkA or TrkC. *J Neurosci Res.* 80: 18-28, 2005

Evangelopoulos ME, **Weis J**, **Krüttgen A**. Signalling pathways leading to neuroblastoma differentiation after serum withdrawal: HDL blocks neuroblastoma differentiation by inhibition of EGFR. *Oncogene.* 24: 3309-18, 2005

Di Martino E, **Sellhaus B**, Haensel J, Schlegel JG, Westhofen M, Prescher A. Fallopian canal dehiscences: a survey of clinical and anatomical findings. *Eur Arch Otorhinolaryngol.* 262: 120-6, 2005

Cordoba F, Braathen LR, Weissenberger J, Vallan C, Kato M, Nakashima I, **Weis J**, von Felbert V. 5 aminolaevulinic acid photodynamic therapy in a transgenic mouse model of skin melanoma. *Exp Dermatol.* 14: 429-37, 2005

Buss A, **Sellhaus B**, Wolmsley A, Noth J, Schwab ME, Brook GA. Expression pattern of NOGO-A protein in the human nervous system. *Acta Neuropathol (Berl).* 110: 113-9, 2005

Hans FJ, Reinges MH, **Nolte K**, Reipke P, Krings T. Primary lymphoma of the skull base. *Neuroradiology.* 47: 539-42, 2005

Saxena S, Bucci C, **Weis J**, **Krüttgen A**. The small GTPase Rab7 controls the endosomal trafficking and neuritogenic signalling of the NGF receptor TrkA. *J Neurosci*. 25:10930-40, 2005

Senderek J, Krieger M, Stendel C, Bergmann C, Moser M, Breitbach-Faller N, Rudnik-Schöneborn S, Blaschek A, Wolf NI, Harting I, North K, Smith J, Muntoni F, Brockington M, Quijano-Roy S, Renault F, Herrmann R, Hendershot LM, **Schröder JM**, Lochmüller H, Topaloglu H, Voit T, **Weis J**, Ebinger F, Zerres K. Mutations in SIL1 cause Marinesco Sjögren syndrome, a cerebellar ataxia with cataract and myopathy. *Nature Genetics*. 37:1312-14, 2005

Schröder JM, Züchner S, Dichgans M, Nagy Z, Molnar MJ. Peripheral nerve and skeletal muscle involvement in CADASIL. *Acta Neuropathol (Berl)*. 110: 587-99, 2005

Goebel HH, Kiefer R, Pongratz D, **Schröder JM**, von Moers A. Indikation und Qualifikationskriterien für die Biopsie und präparative Aufarbeitung von Muskel- und Nervengewebe. *Nervenheilkunde*. 24: 933-35, 2005

Block F, **Weis J**: Periphere Neuropathie, Kap. 17, S. 303 – 328. In: Block F, Prüter C (Hrsg.) Medikamentös induzierte neurologische und psychiatrische Störungen, *Springer Medizinverlag Heidelberg*, 2006

Block R, **Weis J**: Myopathie, Kap. 19, S. 343 – 357. In: Block F, Prüter C (Hrsg.) Medikamentös induzierte neurologische und psychiatrische Störungen, *Springer Medizinverlag Heidelberg*, 2006

Publications 2004

Züchner S, Vorgerd M, Sindern E, **Schröder JM**. The novel neurofilament light (NEFL) mutation Glu397Lys is associated with a clinically and morphologically heterogeneous type of Charcot-Marie Tooth neuropathy. *Neuromuscul Disord*. 14: 147-57, 2004

Züchner S, Mersiyanova IV, Muglia M, Bissar-Tadmouri N, Rochelle J, Dadali EL, Zappia M, Nelis E, Patitucci A, Senderek J, Parman Y, Evgrafov O, Jonghe PD, Takahashi Y, Tsuji S, Pericak-Vance MA, Quattrone A, Battaloglu E, Polyakov AV, Timmerman V, **Schröder JM**, Vance JM. Mutations in the mitochondrial GTPase mitofusin 2 cause Charcot-Marie-Tooth neuropathy type 2A. *Nature Genetics*. 36: 449-51, 2004

Wu YJ, **Krüttgen A**, Moller JC, Shine D, Chan JR, Shooter EM, Cosgaya JM. Nerve growth factor, brain-derived neurotrophic factor, and neurotrophin-3 are sorted to dense-core vesicles and released via the regulated pathway in primary rat cortical neurons. *J Neurosci Res*. 75: 825-34, 2004

Weissenberger J, Loeffler S, Kappeler A, Kopf M, Lukes A, Afanasieva TA, Aguzzi A, **Weis J**: IL-6 is required for glioma development in a mouse model. *Oncogene*. 23: 3308-16, 2004

Weigel R, Senn P, **Weis J**, Krauss JK. Severe complications after intrathecal methotrexate (MTX) for treatment of primary central nervous system lymphoma (PCNSL). *Clin Neurol Neurosurg*. 106: 82-7, 2004

Vielhaber S, Feistner H, **Weis J**, Kreuder J, Sailer M, **Schröder JM**, Kunz WS. Primary carnitine deficiency: adult onset lipid storage myopathy with a mild clinical course. *J Clin Neurosci*. 11(8): 919-24, 2004

Saxena S, Howe CL, Cosgaya JM, Hu M, **Weis J**, **Krüttgen A**. Differences in the surface binding and

endocytosis of neurotrophins by p75NTR. *Mol Cell Neurosci*. 26: 292-307, 2004

Rohde V, Reinacher P, Patz E, **Sellhaus B**, Gilsbach JM. Spinal cord compression by a cervical oseteocartilaginous exostosis: surgical strategy aspects. *Z Orthop Grenzgeb*. 142: 179-83, 2004

Mariani L, Siegenthaler P, Guzman R, Friedrich D, Fathi AR, Ozdoba C, **Weis J**, Ballinari P, Seiler RW. The impact of tumour volume and surgery on the outcome of adults with supratentorial WHO grade II astrocytic tumors. *Acta Neurochir (Wien)*. 146: 441-48, 2004

Mariani L, Schaller B, **Weis J**, Ozdoba C, Seiler RW. Esthesioneuroblastoma of the pituitary gland: a clinicopathological entity? Case report and review of the literature. *J Neurosurg*. 101: 1049-52, 2004

Evangelopoulos ME, **Weis J**, **Krüttgen A**. Neurotrophin effects on neuroblastoma cells: Correlation with Trk and p75NTR expression and influence of Trk receptor bodies. *J NeuroOncol*. 66: 101-10, 2004

De Paepe B, **Schröder JM**, Martin JJ, Racz GZ, De Bleecker JL. Localization of the alpha-chemokine SDF-1 and its receptor CXCR4 in idiopathic inflammatory myopathies. *Neuromuscul Disord*. 14: 265-73, 2004

Christiansen S, Demircan L, Kwant PB, Akdis M, Rex S, Buhre W, Langebartels G, Kuruc N, **Nikolin S**, Reul H, Autschbach R. Experimental testing of a new left ventricular assist device - the microdiagonal blood pump. *Asaio J*. 50: 200-4, 2004

Chen L, Schaerer M, Lu ZH, Lang D, Joncourt F, **Weis J**, Fritschi J, Kappeler L, Gallati S, Sigel E, Burgunder JM. Exon 17 skipping in CLCN1 leads to recessive myotonia congenita. *Muscle Nerve*. 29: 670-6, 2004

Buss A, Assmus A, Weidemann J, **Sellhaus B**, Lorenzen J, Block F. Diagnosis of an initial infratentorial central nervous system B-cell lymphoma during prolonged cortisone medication. *Nervenarzt*. 75: 1217-21, 2004

Bissar-Tadmouri N, Nelis E, **Züchner S**, Parman Y, Deymeer F, Serdaroglu P, De Jonghe P, Van Gerwen V, Timmerman V, **Schröder JM**, Battaloglu E. Absence of KIF1B mutation in a large Turkish CMT2A family suggests involvement of a second gene. *Neurology*. 62: 1522-5, 2004

Bahm J, Becker M, Disselhorst-Klug C, Williams S, Meinecke L, Müller H, **Sellhaus B**, **Schröder JM**, Rau G. Surgical Strategy in Obstetric Brachial Plexus Palsy: The Aachen Experience. *Seminars in Plastic Surgery* 18: 285-99, 2004

Korinth MC, Kapser A, **Nolte K**, Gilsbach JM. Cervical diastematomyelia associated with an intradural epidermoid cyst between the hemicords and multiple vertebral body anomalies. *Pediatr Neurosurg*. 40: 253-6, 2004

De Paepe B, Racz GZ, **Schröder JM**, De Bleecker JL. Expression and distribution of the nitric oxide synthases in idiopathic inflammatory myopathies. *Acta Neuropathol (Berl)*. 108: 37-42, 2004

Schröder JM, Durling H, Laing N. Actin myopathy with nemaline bodies, intranuclear rods, and a heterozygous mutation in ACTA1 (Asp154Asn). *Acta Neuropathol (Berl)*. 108: 250-56, 2004

Schröder JM, Hackel V, Wanders RJ, Gohlich-Ratmann G, Voit T. Optico-cochleo-dentate degeneration associated with severe peripheral neuropathy and caused by peroxisomal D-bifunctional protein deficiency. *Acta Neuropathol (Berl)*. 108: 154-67, 2004

Gossrau G, Gestrich B, Koch R, Wunderlich C, **Schröder JM**, Schroeder S, Reichmann H, Lampe JB. Apolipoprotein E and alpha-1-antichymotrypsin polymorphisms in sporadic inclusion body myositis. *Eur Neurol.* 51: 215-20, 2004